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School of Education



The experiences of people with rare
syndromes and sensory impairments
in hospitals and clinics

Dr Liz Ellis, (University of Birmingham) Lucy
Keenan (Sense), and Dr Liz Hodges (University of
Birmingham)

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2 Glossary and abbreviations

2.1 Glossary

Chorionic Villus Sampling – a medical test which can identify certain abnormalities in an foetus by taking a sample of the placenta.

Ciliopathy – a disease of the *cilia* - 'hairs' which perceive sensory function. Alström, Bardet-Biedl, and Usher are ciliopathy conditions.

Communication professional – a person whose job is to act as interpreter or communication intermediary, in this case for people who are deaf or deafblind and use communication methods other than speech

Cryotherapy – the use of low temperature in medical treatment. In this report it refers especially to the use of a cold probe to freeze a detached or detaching retina in place.

Deafblind manual – a method of spelling out messages on a deafblind person's hand, letter by letter.

Hospital Passport – a document listing a patient's needs, communication methods, likes and dislikes and sometimes more, for someone who cannot communicate well themselves or who needs to show it to many people.

Intervenor – an individual working in education or social care to support the needs of a congenitally deafblind person and give them access to their setting

Makaton – a sign language vocabulary system especially designed for people with learning disability

Retinal dilation – a procedure using eye drops to make the pupil expand so that the retina can more easily be seen by a doctor. As the retina is forced to expand, more light gets in and patients are very affected by glare and cannot see clearly.

Retinal pigment epithelium – the retinal layer that nourishes retinal visual cells

Slit lamp – a medical instrument using a bright beam of light to examine the eye

2.2 Abbreviations

BBS – Bardet-Biedl syndrome

BCH – Birmingham Children's Hospital

BSL – British Sign Language

CRS – Congenital Rubella Syndrome

GOSH – Great Ormond Street Hospital

IRD – Infantile Refsum Disease

NF 2 – Neurofibromatosis type 2

QE - Queen Elizabeth Hospital Birmingham

SSE – Sign Supported English

3 Executive Summary

This study was born from informal discussions with people with rare syndromes (particularly those with sensory impairments) who were describing difficulties they were having as **patients**. In many cases they were already carrying a large burden of problems; they had multiple, complex individual symptoms, and they were often aware their conditions could or would deteriorate; they were dealing with a world unfriendly to people with sensory impairments.

The research study was therefore set up to investigate the following issues;

- How do people with sensory impairments (and their families/carers) experience their attendance at clinics in relation to their rare syndromes?
- What are the factors in relation to clinic type, environment, and attitude that affect this experience?
- What makes the delivery of services efficient and effective for people with sensory impairments and their families, across and between clinics and what factors might inhibit this?

3.1 What is good practice?

These Good Practice Guidelines are presented first, as they summarise what was learnt from the study. However, they were compiled from the information gathered from interviews, environmental data and accompanied visits. They are thus drawn from patient experience as expressed to us through the study, and emerged from the data, rather than being a pre-existing measure for the hospitals we visited and talked about.

These guidelines fall under three broad headings; Good practice in the environment, Good practice by staff, and Good practice in clinical situations.

They are divided into two sections, the first section includes items which might require authorisation, expenditure, systems reorganisation or other

large scale thought and work. The second section however requires much less in terms of infrastructure, although it might involve training, and is more concerned with simple, and inexpensive changes which can be undertaken by people who are prepared to make an effort.

Good practice in the environment

Policy strategies

- Buildings are easy to navigate, using colour coding of areas, clear and consistent signage, multiple formats of information
- There is good even lighting throughout corridors and in cafés and toilets
- There is at least clarity about car parking – such as patients only having to pay for what they have consumed (such as payment on exit)
- Department names are kept the same throughout the site (so not both “eye clinic” and “ophthalmology”)
- Toilets for people who need to lay down to be changed are made available
- All consulting rooms to have four walls and a solid door

Operational strategies

- Reception staff should offer directions and support for wayfinding on arrival
- Lights are kept on and additional lighting is provided in dark areas
- More play/activity equipment is provided in waiting areas, particularly thinking about the needs of older children, and also children who require specific sensory toys. TVs with subtitles and signing, and wifi are made available.
- Deliberate and clear paths are kept across waiting spaces – perhaps marked with tape/paint as clear paths
- Toilet leaflets are provided - simple directions for how toilets work, which are offered to patients (see Nina in 8.2.4)

Good practice in staffing

Policy strategies

- All staff, including reception and nursing staff, are trained in issues around sensory impairment, communication techniques and guiding
- There is a review and assessment policy for each hospital which includes a walk through and overview of procedures and clinics – such as 15 Steps, with a focus on sensory impairment issues
- Liaison personnel are provided; non-medical staff who can point patients in the right direction for both medical information, procedural information, and further support
- Training for staff in understanding the issues of dilation (vision reduction) for all patients, but particularly those who are deaf/ have a hearing impairment

Operational strategies

- Use of a sticker or colour-coded protocol or similar which outlines patients' communication and information needs and which all staff **read**. This is now a part of the Accessible Information Standard and will need to be implemented by July 2016
- All staff, reception and clinical, regularly check communication needs - 'Is that communication OK for you? Can I ask to you to repeat that so I know you understand?'
- Time is always given for genuine listening to patient – for those with rare syndromes only they can know the individuality of their conditions
- All staff always introduce themselves, clearly, including their job title, and this information is available in written (print, email, braille) format both before, and after appointments
- Staff give patients time, understanding that patients may have travelled a long way and have waited a long time
- Staff look at patients when talking, do not cover their mouths and pay full attention to patients during discussion
- Staff talk to patients, not interpreters or companions

- Staff ask if patients would like them to read information leaflets/forms/documents to them, and assist in filling out forms
- A culture of asking three key questions;
 - How can I help?
 - Am I getting this right?
 - What else can I do?

Good practice in clinic practice; appointments and procedures

Policy strategies

- Multiple formats of leaflets/information are always available (large print, audio, braille, electronic) and given to patients rather than leaving them to be noticed by patients
- Co-ordinated appointments are offered to minimise pressure on patients and families – either for families (syndromes often run in families) or multiple conditions (so that audiology and cardiology are carried out on the same day, or are on the same day for siblings)
- Procedures requiring anaesthesia are grouped to minimise the number of anaesthetics given
- Formal arrangements are made for patients to be able to meet by having ‘syndrome’ days for patient appointments
- Informal arrangements are encouraged for patients to meet each other – perhaps co-ordinated on social media, but a room is provided
- Loop systems are always available, working (checked) and used
- A quiet, private room is available for discussion with the receptionist, clinician or other

Operational strategies

- Appointment and post appointment information is available in accessible formats, for individual needs; always checking, is this format still Ok for you?

- Appointment and post appointment information is always available in digital formats if this is preferred
- Patients are given clear advance information about what the appointment is for; and what might happen as a result of it (patients were not always clear and some are juggling so many appointments, they have to prioritise)
- A plan of what exactly will happen at appointment or clinic is provided in advance, with who they will see, to minimise confusion
- Text messages are used to aid communication – e.g. to confirm appointments or tell people when to return from the café or outside if clinics are running late
- Information is promptly given in appropriate formats about waiting times; reasons for waiting; text messages are sent about delays
- There is a clear system for ensuring that people know when they are called to an appointment once in clinic, including approaching people directly if they need this
- Staff willingly talk to patients if they have difficulty with automated systems (e.g. booking in machines)
- Specialist clinics (e.g. ophthalmology, audiology) understand the needs of their own patients- e.g. vision impairment or hearing impairment
- There are smaller areas (with screens or similar) for children who find noise and bustle difficult
- Staff are ready to guide people to rooms when they are called and will ensure that someone will take them back
- Staff are always willing to provide, afterwards, written feedback on what was said in an appointment, in appropriate formats, especially digital information
- Named contact for further information following the clinic visit

3.2 Patients, clinics and syndromes

We focused on six particular syndromes which frequently cause dual sensory impairments and which are all rare (fewer than one in 2,000 people have them). Amongst the considerations which led us to these groups were; that they all cause sensory impairments and frequently dual sensory impairments; that all had particular clinics or practitioners who focused on them; that several of them were ciliopathies; that there was a patient identity associated with conditions, related to, for example, the existence of a support group.

The syndromes we thus included were; Alström syndrome; Bardet-Biedl syndrome; CHARGE syndrome; Stickler Syndrome; Usher syndrome and Wolfram syndrome. We also included the experiences of 5 people with syndromes that were different to these six main syndromes.

Wolfram syndrome's key factors are diabetes insipidus, diabetes mellitus, optic atrophy and deafness. It is a degenerative disorder. It affects 1 in 770,000 people in the UK.

Alström syndrome affects the whole body, including with rod-cone retinal dystrophy, sensorineural hearing loss, obesity, insulin resistance, and type 2 diabetes mellitus, as well as a range of other issues. It is a life-limiting disorder, which might affect 100 people in the UK .

Stickler syndrome is a group of conditions which affect connective tissue (collagen). Stickler is characterised by a distinctive facial appearance, cleft palate, eye problems, hearing loss, and joint problems. It affects about 1 in 8,000 people in the UK

CHARGE syndrome's principal factors are with the eye, choanal atresia (when the nasal passages are blocked by bone or tissue), cranial nerve anomalies, and ear anomalies. They may have learning delay. It affects about 1 in 12,000 people in the UK.

Usher syndrome (type 1, 2 and 3) is principally characterised by sensorineural deafness and progressive vision loss due to Retinitis Pigmentosa (RP). Initially peripheral vision loss occurs which is described as 'tunnel vision'. The exact number of people affected is unknown but it could be as high as 1 in 7,000 people.

Bardet-Biedl syndrome's main features are retinal degeneration, extra fingers and/or toes and obesity. Diabetes mellitus is also common. It affects about 1 in 160,000 people in the UK.

This study looked predominantly at three kinds of clinics;

Specialised holistic services; clinics provided yearly with a range of specialisms involved for a particular syndrome (Alström, Wolfram and Bardet-Biedl each have these type of clinics).

Condition specific clinics; a clinic in which a consultant and the team will focus on a particular condition (Stickler, Usher and CHARGE syndrome have these kind of clinics)

General clinics; clinics which are neither of the above but are still often highly specialist.

3.3 Methodology

In the course of this project, over 20 months, we gathered information from 52 participants (either people with rare syndromes and sensory impairments or their families), from all over England, representing people from 14 months to 83 years old. We have collected many hours of interview data, including some in BSL, and attended 5 hospitals, incorporating 8 clinics, to see how things work in practice, where the clinics are situated, and sometimes alongside the people attending.

The study incorporated a number of information gathering tools

Interview with patients; initially patients with one of the six syndromes who attended specialist clinics were interviewed; later this expanded to other people with dual sensory impairments and with some other conditions who attended a range of clinics. The interviews investigated travel and mobility to and within the hospital, the information provided before and after the clinics, and communication from receptionists, clinical staff and others.

Environmental audits; these were carried out in five hospitals to provide additional data for comparison. They examined patient areas such as corridors, waiting areas, toilets and cafés, and clinic rooms.

Accompanied visits; we also accompanied five patients on their visits through hospitals, to gain some insight into how long procedures took, what communication methods staff used, how aware staff were of the needs of people with sensory impairments, the time given to communicate, including reading papers, and more.

The study gained ethical approval from the NHS (NREC) and from each of the participating hospitals. Patients and their families were asked for their consent, using appropriate formats considering age, communication method, and ability. 52 people were interviewed and told us of their experiences of rare syndromes, clinics, and sensory impairments; of these, 42 were people who have a rare syndrome, and the remaining 10 were from parents/guardians of people with a rare syndromes speaking about the experiences of clinics for the people they care for and in their own role as a parent or carer.

3.4 The patient experience;

From the exploration of the experiences of participants we analysed the data gain from the three research tools to draw out points in relation to key moments in the patient's journey through the clinic; appointments, in the clinic, the consultation, and after the clinic.

3.4.1 Appointment

Patients talked about issues around getting an appointment, and the different routes to this depending on the kind of clinic they were attending. They considered specialist clinics and clinicians to be very valuable, although distances to clinics and the length of time between referrals could be a problem. The process of making an appointment was not always clear to the participants; and they did not feel their individual needs (such as sensory impairment and communication requirements) were always taken into account. The complexities of their conditions along with juggling personal factors (for example childcare, work commitments, school timetables) meant that participants sometimes had to balance how many appointments they could go to. Most participants who were offered multiple appointments in one day (at specialist clinics) preferred this, and also preferred additional

appointments where possible to be dealt with at school, over the telephone, or by a visit to the home.

3.4.2 At the hospital

Difficulties with travel to hospital are exacerbated by sensory impairment, and the rarity of their conditions also meant they often had to travel a long way to specialist facilities.

In looking across different hospitals, some had good lighting and minimised glare, some had good colour schemes which identified areas and furniture, some had clear, good sized signs, some avoided clutter in areas where people were moving, but none of the hospitals managed all of these. Once within the hospital, cafés could be expensive and sometimes they were a long way from the clinic, they were not well lit, and they did not provide for sensory needs. Toilet facilities, obviously important when people are waiting for some time were sometimes easy to access, but others found that sensory issues, made it difficult for them to be independent.

3.4.3 At the clinic

Patients talked about their experience of reception areas, waiting and the role of specialist co-ordinators. Most frequently they talked about the key needs for face-to-face communication, staff looking while speaking, speaking up and speaking to the patient rather than a companion.

Waiting is central to the patients' experience of hospitals. Participants appreciated information about how long they might need to wait, and why, in appropriate formats. Consideration to individual needs and preferences such as appropriate activities for children of different ages, TV with subtitles, or wifi would also be appreciated. Participants worried **a lot** about not hearing their names called for their appointment. Some participants appreciate (or would appreciate) the opportunity to meet other people with their syndrome, but others would prefer this to be through network groups. Clinic co-ordinators, where they existed, performed a vital role as a friendly face, in helping with logistics, and as a point of contact between clinic visits.

3.4.4 Consultation

In the consultation, participants experienced different levels of communication, with some clinical staff being very aware of individual needs, whilst others were not. Unfortunately, poor communication skills were seen and described far more often than good ones. Participants whose sight was very impaired, were not told what was happening during appointments, such as silence while people wrote notes. Participants were frustrated by staff talking to their companions rather than them and by inadequate or inconsistent provision of communication support (where needed).

Participants needed information which sighted/hearing patients did not, such as being able to touch equipment and they needed information in appropriate text formats (large print, braille, digital). Hearing impaired patients needed spoken information backed up later, such as emailed reports on the clinic visits to read in their own time. Participants also felt strongly that staff should be honest about procedures which hurt or were uncomfortable or inconvenient. Clinics were usually well lit, but sometimes unnecessarily noisy, because doors were left open, or some areas were only bounded by curtains.

3.4.5 Beyond the clinic

Participants frequently felt they needed some support beyond the appointment, both from the hospital and from other organisations, but they often felt this was lacking. Where there were clinic co-ordinators pathways for support after the clinic were clear, but otherwise they often did not know who to contact. Referrals to support organisations such as Sense would have been welcomed by many, and where they were made, they felt supported. Many participants said they would like to be in touch with others with the same syndrome, and where they were, they found this useful.

3.4.6 Additional points

Some points about staying in hospital and GP clinics were also raised by participants. Hospital stays were often very difficult for patients with sensory impairments, with hospital staff not understanding their communication and mobility needs. For children and their parents, such

stays were very stressful. In visiting GP surgeries, participants raised again the issues of inaccessible information; and not being able to get consultations in appropriate formats. Where this was working well, they had built up relationships by seeing the same GP over some time.

3.5 People with sensory impairments and rare syndromes

For people who had dual sensory impairments and rare syndromes, they found the key areas of mobility, communication and access to information difficult in their hospital visits as in all other areas of life. Their difficulties in each of these areas were in some cases the same as other people's (for example, parking in relation to mobility) but sometimes exacerbated by sensory impairment (for example, signage which they could not see, and they were not able to ask their way because they could not hear the answer) and sometimes was different to that of people who do not have sensory impairments (for example, managing bathroom furniture with no contrast e.g. all white). For each area, some simple strategies would potentially help considerably, for instance, all staff being trained to ask 'Is this communication OK for you?' could allow patients to ask someone to speak up, face them, or read documents to them.

For patients with these rare syndromes, the issues relating to their lives, and their medical treatments all involved connectivity, multiplicity, rarity, and individuality. They needed their services linked together and each aware of what others were doing. They needed clinical staff to understand that their conditions were not just single issues, but each one affected all the others. They knew their conditions were rare and they both appreciated staff who did understand something about it but also those who were prepared to listen to patients.

But, and as an important coda to all the above, these patients were above all, people. They were individuals in the nature of their syndromes and sensory impairments (no two the same) but as well as being patients, participants spoke of being sons and daughters, pupils, musicians, cooks, friends, husbands and wives, mothers, campaigners, sportsmen, artists, and many more identifiers. We would not wish to

forget this rounded perspective in our descriptions of the small element of their lives that was as “patients”.

4 Introduction

4.1 The origins of this study

This study was born from informal discussions with people with rare syndromes (particularly those with sensory impairments) who were describing difficulties they were having as patients. In many cases they were already carrying a large burden of problems; they had multiple, complex individual symptoms, and they were often aware their conditions could or would deteriorate; they were dealing with a world unfriendly to people with sensory impairments. They had chronic health care needs and sometimes also acute ones; these were complicated to manage and they needed their health care to help them rather than make things more difficult. For some of these conditions there were specialist clinics, for others specialist centres, and these were often very involved in research, but it was not always research that focused on the immediate, direct needs of the patients.

These individuals wondered if things could be better at the hospitals they visited. Sense, with the support of specialist syndrome groups such as the Ciliopathy Alliance, and others, decided to try to discover what was working well, and what could be improved and so funded this research project.

This project was focused on three issues; people with rare syndromes; the sensory impairments often caused by these syndromes; and the clinics they attended. Of most interest was the role of the specialist clinics, those which are particularly named to certain rare syndromes and are nationally commissioned. The study included other hospitals who were trying to do something similar. However, it was widened to include a range of clinics and outpatient services for people with rare syndromes and sensory impairments.

In looking at the research literature, there was little focused on the needs of people with *dual sensory impairments* in hospitals, and little which looked at the whole experience of people with complex conditions, rather than focusing on the different areas of need. We hoped to address these issues.

4.2 Research questions

The research study was therefore set up to investigate the following issues;

- How do people with sensory impairments (and their families/carers) experience their attendance at clinics in relation to their rare syndromes?
- What are the factors in relation to clinic type, environment, and attitude that affect this experience?
- What makes the delivery of services efficient and effective for people with sensory impairments and their families, across and between clinics and what factors might inhibit this?

To do this, a qualitative study of people attending clinics was devised, drawing on their experiences through their voice in interviews, through environmental audits of hospitals and clinics, and through accompanying patients on visits. Patient stories are recognised as a powerful way of encouraging medical staff to understand what receiving medical care is like – for the patient. More details on participants, methods and design are included in section 7.

4.3 Research background and protocol

The researchers for this study were experienced in research and also in working with and communicating with people with dual sensory impairments. With this knowledge and background they looked at situations through the lens of their understanding about the needs of people with sensory impairments to inform the research tools and design. The researchers believe that access to provision is important, and achievable and we want to share this belief with the clinical staff who worked with the patients and to improve provision for the patients themselves.

This report is therefore intended to be of interest to the following groups

- Medical practitioners interested in the patient journey for people with these rare syndromes
- Specialist and campaigning organisations for people with dual sensory impairments
- Specialist groups related to the rare syndromes represented
- Individuals with and the families of those with rare syndromes or dual sensory impairments or both

4.4 Research outline

In the course of this project, over 20 months, we gathered information from 52 participants (either people with rare syndromes and sensory impairments or their families) with 10 rare conditions, from all over England, representing people from 14 months to 83 years old. We have collected many hours of interview data, including some in BSL, and attended 5 hospitals, incorporating 8 clinics, to see how things work in practice, where the clinics are situated, and sometimes alongside the people attending.

In this report we have first detailed the literature that underlies the concept of the 'patient experience' and then the literature about the experiences of patients with sensory impairments and rare syndromes. We then look in more detail at the rare syndromes themselves and at the kind of clinics people attend. The methodology, methods and ethics of the research are then outlined. In our analysis of the data collected, we have followed a typical patient journey through a clinic, starting with appointments, then travel, then being at the hospital, through the clinic itself, the consultation and then what happens afterwards. We have followed this with a discussion of themes and an overview of good practice.

5 Literature review

To support the study we first looked at literature about patients' experiences in hospitals, clinics and about hospital design. We searched a number of databases but it was very hard indeed to find material about the experiences of people with sensory impairments in clinics. While there was considerable literature about patient experiences, much of this was a response to clinical situations, in terms of treatment outcomes and patient care. This was not what we were looking at.

5.1 How can we study the *Patient experience*?

There is a body of literature related to the patient experience, and exploring what this is. There are many articles devoted to the patient experience within the medical literature. The patient experience has become a much more significant factor in the evaluation of health care in more recent years. As stated by Wolf et al (2014) it is about more than the 'results of surveys' and deeper than 'satisfaction alone'. It is much more related to the efficacy of health care in meeting the needs of individuals, with their individuality, their needs, expectations and lives. As such, the gathering of any patient experience data is just that; it represents individual patients think and feel, and cannot be simply analysed in terms of scores or numbers. Individuals view the same service differently. For some studies, the patient experience was summed up within the use of an

“established set of standards, and a set of measurable indicators;”
(LaVela and Gallan, 2014 pg 29)

but others go beyond this to explore what it is like to be a patient. This literature informed our study protocols and is the established background for our study.

LaVela and Gallan (2014) outline the importance of using a range of different approaches to gathering information about the patient experience, including what they call 'ethnographic' approaches, such as

unobtrusive observations and patient journey mapping. These kinds of approaches they suggest, using mixed methods, are able to give an 'in depth understanding of patient experiences' which is not achieved by numerical or rating measures alone. A more open approach to finding out what is important enables an understanding of the real time experiences of people as patients. Amongst other methods they suggest *Shadowing* and *Guided tours*. In Guided Tours, patients take a researcher through their hospital visit, explaining the factors which are important to them, including their 'thoughts and feelings'. As they describe;

"This method is able to capture a multi-sensory (e.g. sights, sounds) insight into the patient experience" (pg 33)

In similar ways, others have looked at listening to patient voices, not within the context simply of treatment or clinical outcomes but as alongside the patient. Robert et al (2011) reported on evidence about patients' experiences in the NHS and emphasise the importance of the patient's story, as having the most significant effect on staff. As Luxford and Sutton (2014) say this kind of narrative

'can provide insight into expectations of care, often in a manner that health care professionals may find more engaging.' (pg 21).

Robert et al and Luxford and Sutton all emphasise the importance of patient stories which, while they do not give scores or numerical values, may have greater actual influence on the practice of medical staff.

The *15 Steps Challenge* was initiated from a remark by a parent,

'I can tell what kind of care my daughter is going to get within 15 steps of walking on to a ward' (Thomas and Clarke 2015 pg 18)

and has been adopted as a self evaluation strategy for hospital departments, including outpatients. It is not based on outcomes but instead looks at questions about *Welcoming and caring, Calmness and organisation, and Information*.

5.2 What patients have reported about experience

Patients are often asked about their experiences in NHS clinics (and in-patient treatment; sometimes the two are not separated). There are many measures, although most are ratings, but only a few are picked out here, those considered especially relevant to this study.

A study in Scotland (Coates-Dutton and Cunningham-Burley 2009) looked at children's experiences through focus groups and among the many factors they reported were

- The importance of appropriate toys in waiting rooms
- Smell

Toys which meet children's needs (having something to do) was a very important issue for the children. There were toys, but they were often for children of only certain ages, usually babies and toddlers, and the children were not able to control what was on the TV. A very significant number of the children remarked on the smell of hospitals and how they disliked it. This might be a particularly significant feature of hospitals for children with dual sensory impairments.

As part of Robert et al's (2009) investigation into the patient experience, they discussed the importance of making such evaluations available to all clients, including those with learning disabilities. They report on the Hertfordshire NHS Trust's innovation in using easy read versions of patient evaluation questionnaires, with pictures and simplified response scales.

A study by Cunnet (2010) on '*What does good look like*' mentioned communication as the most important factor in patients feeling their needs were well met, while the environment was much less mentioned, and mostly in relation to cleanliness, and parking. Flexibility in meeting the needs of individuals was also raised as an important factor.

Mercieca, et al (2014) also looked at the patient journey through a rheumatology clinic and discussed key factors such as inappropriate waiting areas, with no space for wheelchairs, car parking, and lack of organisation at reception, where staff are frequently multi-tasking and cannot pay attention to individuals.

Reeves and Bruster (2009) in Scotland and Boyd (2007) in England (both cited in Robert et al 2011) report on the ten most important and ten least important factors in patient care, from patient reported surveys. These are of interest to this study both in terms of what they say, but more significantly in terms of the potential shortcomings of such studies in relation to the sensory impaired patients on whom this study is focused. For example, among the ten *least important* in both studies was 'being able to get access to an interpreter'. It must be assumed however that this is largely a factor related to the number of people asked, that, assuming it was a large number, only a relatively small number of them would have needed an interpreter, therefore it appears unimportant. For the individual who *does need* an interpreter, it is, we can assume, crucial. Without an interpreter, for a BSL user for example, those items raised as in the top ten most important to most people 'I get a clear explanation' and 'risks and benefits are explained to me in a way I can understand' would hardly be achievable. Likewise, 'I am not bothered by noise during the day' was amongst least important items, but again, could be very important for effective communication with someone with impaired hearing. Although patients with sensory impairments may visit hospitals more often, they may be less represented in such wide ranging studies, and thus become a low priority for hospital trusts seeking change.

5.3 Patient experience – looking at the environment

There are many documents which provide standards for evaluations for hospital environments, in a range of details and with many factors. A number of these highlight good practice in hospital environments which are likely to be helpful to visually impaired people. NHS Wales (2004) reports on, among many other factors, the importance of clear way marking, through using colour schemes, and good labelling at named areas when they arrive, the value of good signage, especially when it is going to be seen from a distance, but also on issues such as handrails contrasting with walls, vertical edges of doors being in contrast to the rest, and obstacles standing out well from their surroundings, which are more specific to visually impaired people.

Of particular significance perhaps are the *Patient-Led Assessments of the Care Environment (PLACE)* for the NHS which cover a number of areas, one being Outpatients and another External Areas (which includes parking and route finding). These assessments are intended to be carried out annually and the evaluation looks at the environment and not at clinical care standards. They are obligatory and reports on them are publically made (NHS 2013).

The outpatient assessment includes cleanliness, condition, access, a dementia assessment, staff appearance, waiting, privacy and wellbeing (NHS PLACE Outpatients 2013). Interestingly the dementia assessment includes many features which would significantly benefit patients with sensory impairments, for example

- toilet seats, handles and rails contrast with the wall and floor
- flooring is noise reducing/absorbent and contrasts with walls
- signs have large, easily readable text
- a large clock face visible in all areas

Likewise, Hignett (2012) reported on signage, flooring and lighting to enhance independence in older, frail and confused patients, all factors which would also increase independence in people with sensory impairments.

The access list in the PLACE assessment includes

- a hearing loop or similar portable system at reception desk
- an audible/verbal appointment alert for visually impaired people, and a visual appointment alert system for hearing impaired people (both of these are the only unscored questions in the evaluation, but they are still included)

The general appearance list includes even natural light, sufficient to make areas bright and easy to walk through.

The external assessment (NHS PLACE External 2013) includes

- clearly marked, well lit and safe journey from a parking area
- trees trimmed so as not to provide an overhang hazard
- clear signage

As well as the physical environment, the communication environment is addressed in the National Institute for Health and Clinical Excellence guidance on patient experience (NIHCE 2012) which includes the importance of using “the most effective way of communicating with each patient” (pg 79) and providing information accessibly. It discusses the use of

‘pictures, symbols, large print, Braille, different languages, sign language or communication aids, or involving an interpreter, a patient advocate or family members’ (pg 79).

It recognises the possible cost implications of this but suggests that in fact, using appropriate communication might result in fewer return visits and thus ultimately save money.

5.4 Patient experience; patients with sensory impairments and rare syndromes.

Finally in this literature review, sources related particularly to the patient experience for those with sensory impairments and/or rare syndromes are presented.

5.4.1 Patients who have hearing impairment

Patients who are D/deaf or have hearing loss report particular problems with communication. Patients using BSL need interpreters (e.g. McAleer 2006), but not all hearing impaired patients need this. Reported in the literature in many articles reviewed was the issue of knowing when patients are ‘called’ to their appointments. Barnett (2002) for example discussed how someone struggled to care for children while also trying to look at reception staff constantly. Ubido (2002) also talks about patients with hearing impairment (deafness) being made to feel ‘inadequate and stupid’ because they did not hear when they were

called. Hines (2000) reported that only 6% of respondents to his study (who were all lip-reading students, so used spoken communication, not BSL) said that the communication skills of staff were good. 64% indicated that they considered hospital staff were trying their best, but did not use appropriate methods, while 36% considered they were not even making the effort to communicate. Of course for hearing impaired patients, one route (e.g. having a BSL interpreter) is not sufficient for all users, as individual patients use different, and often multiple routes to communicate (Middleton et al. 2009). For patients with dual sensory impairments the problems are even greater. Palmer, (2015) in an informal blog, outlined the problems of hearing impaired (deaf) people when communicating with health care professionals, giving many examples, such as not knowing what medication to take. He reports that NHS staff were shocked by some of the issues mentioned by the deaf people, they were not aware of what was happening. He notes particularly, that because of their hearing impairment, hearing impaired patients are less likely to complain.

Even in audiological clinics, patients found that staff did not understand the communication needs of hearing impaired (deaf) people (Hines 2000). One patient with hearing impairment, talking about audiology services said

‘You know, audiology should know that deaf people are going to be there’. (Mulla et al., 2014 pg 46)

5.4.2 Patients who have visual impairment

Kopp (2013) as a visually impaired person (and also a nurse), discusses his frustration at the way in which medical staff have not understood his needs. From his perspective, both hospital environments and hospital staff work to make things difficult for visually impaired people. He discusses the need for nursing staff to understand that he needs not to be pushed but guided, that he needs help filling in paperwork, and that

‘one size fits all does not work when you are caring for an individual with a visual impairment’ (Kopp 2013 p 11)

Boyce (2011) and Boyce et al (2014) discussed the importance of wrap around care, and the particular role of the Eye Clinic Liaison Officer, who supports ophthalmology clinics. These professionals, who are not doctors or nurses, nevertheless help to provide a service which meets information needs, provides additional time, and adds to support, thus being cost efficient and saving the time of medical staff. Noticeably, in Douglas et al's (2010) discussion with visually impaired people about their experiences of eye clinics, none of them mentioned anything about accessibility of information –it may have been that they were simply not aware what they were missing (most were recalling the occasion on which they were registered as sight impaired). There may have been leaflets available or help groups advertised in posters, but as the patients could not see them, they cannot report that they missed them. As Douglas (2015, personal communication) then mentioned, in these discussions with participants in which they recall these experiences, such factors as what they might have missed could not be captured. There are many details of these interactions which they might not remember, small things which could have significant effects (such as signage being unreadable). In this way, accompanying patients helps to highlight issues which may not have been recalled.

Rousek and Hallbeck (2010) looked at the environment using simulated visual impairment in relation to wayfinding in hospitals in the USA. This is problematic of course, as the people subjected to simulated conditions did not have the skills, or the disadvantages, of people who were actually visually impaired, however, they reported that most participants had difficulty with signage, which was too small, poorly illuminated or not seen, and encountered many objects in their way, such as benches. The authors recommended that signage should not only be well lit and appropriately lettered but also that there should be a consistent pattern for where it was located.

5.4.3 Patients who have dual sensory impairments

There is little written about patients who are dual sensory impaired apart from by Sense, and authors writing about visually or hearing impaired patients do not generally consider those who have combined visual and hearing impairment.

The Sense guidelines for NHS staff working with dual sensory impaired patients (2013) include:

a checklist for the environment,

- lighting issues,
- contrast and furniture,
- clear signage,
- limiting noisy environments (e.g. not having a radio on),
- glass between patients and reception staff,
- visual displays,
- a hearing loop, *which is used*,

a checklist for staff knowledge,

- using clear speech,
- knowing how to guide,
- longer appointments for people who need more time,
- using a text phone,
- making sure patients have heard their name called

A checklist for information

- recording the communication/information needs of patients
- letters sent in different formats
- a range of ways to contact staff, and patient information in different formats,
- an accessible website,

A booklet (Sense, no date) predominantly about inpatients who are children, mentions the importance of children with dual sensory impairment having access to their hearing aids, and the difficulty they may have in noisy spaces, or spaces with tiles or echoes. It suggests that adjustable blinds are important, and that a severely visually impaired child should be sat near a wall, or curtain to enable them to understand their space. White (2015) gives examples of problems such as not making an appointment for a flu jab because patients with dual sensory impairments could not see the posters telling them to, and not having communication support booked for them. Mulla et al (2014) outline some of the difficulties patients with dual sensory impairments attending audiology clinics had, such as not being able to read the

referral letter, or information that came with their hearing aids, because the font was too small. They also discussed the fact that audiology and ophthalmology departments rarely communicated with each other. Patients felt there was no-one who understood dual sensory impairment, that audiologists would speak to visually impaired clients in the dark and not understand why they could not understand. Sense (2014) gives examples of good care; where a taxi driver takes the patient to reception, the reception staff take her to the waiting room and then the doctor takes her into the appointment. Another participant in the Sense workshop discusses the value of a communicator guide – with the cost met by the NHS. Another patient had her GP suggest he visit her at home so that she could use her computer and ‘deafblind display’ to communicate. But the same workshop mentioned problems with hospital transport that will not allow both a guide dog and a communicator, and with staff assuming that the interpreter required was for BSL when the patient required deafblind manual. Others mentioned difficulties with information in small print, print that could not be scanned, or provided in braille, but too late for the appointment. The complaints procedure, one participant suggested, was not available in accessible formats. The participants in this workshop felt very strongly that they should only have to tell staff about their communication/information needs once and that it should then be written down so that everyone used their communication preferences.

Ellis and Hodges (2013) reported the opinions of people with Usher syndrome who talked about their experience with health care as well as many other things. They also thought that hospitals should understand the individual needs of patients in regard to information, for example, getting letters to them in large print, which for eye clinic appointments they considered should have been routine.

5.4.4 Patients who have rare syndromes

There are of course many patients who have multiple problems, and while rare syndromes are by definition relatively uncommon, having a rare syndrome is not so infrequent, because there are many. Fortin et al (2007) discuss the issue of ‘multi-morbidities’ and suggest that there is little research about how having multiple conditions will affect the patient’s care journey, and there is little understanding of what ‘best

care' for these patients is. As Grut et al (2008) discuss, patients may feel alone, they may not know anyone else with their condition. The patients and their families in this their study in Norway found it very helpful to meet others at specialist clinics. Local services did not understand their conditions. In fact, Limb et al (2010) discuss how at least 25% of patients with rare syndromes attend 3 or 4 different clinics, and 12% attend five. Most had to travel considerable distances for at least some of these. Most attended multiple clinics for different aspects of their conditions but had no specialist centre with a particular interest in their condition. Cadby (2012) reports that many patients with Bardet-Biedl syndrome and those with Neurofibromatosis 2 said they had more than one doctor as a lead in their care, and more of the patients with genetic conditions (which included BBS and NF2) said more frequently than either cancer or transplant patients that information was not passed on between services. 71% of them believed that their GP did not understand their condition (Cadby 2012).

The specialist clinics for Alström, BBS, Wolfram and Stickler (see 6.3.1. below) have service specification standards which talk about patient care. The Alström clinic standards specify a clinical nurse specialist and a 'family centred, one stop clinic', thus providing similar support as to the Eye Clinic Liaison Officer. A wide range of specialists (physician, dietician, physiologist, cardiac technician, phlebotomist), attend, and audiology, cardiology and respiratory clinics are visited and results of tests are to be ready on the second day. It is also specified that literature such as appointments, are sent in appropriate formats including large print and braille. The BBS clinic also outlines a co-ordinated approach with the ophthalmologist, geneticist, endocrinologist, nephrologist, dietician and psychologist all attending. The specification for specialist clinics for Stickler Syndrome outlines that

The day to day management of the associated general refractive, orthoptic, ophthalmological, audiological, musculo-skeletal and cleft issues for each patient will continue to be delivered by local (referring) providers, but the service will continue to offer all affected patients annual MDT review.

while the annual clinic can help to co-ordinate services and ensure that long-term issues are dealt with.

It can be seen then that there are many issues for patients who have rare syndromes and sensory impairments, and while there are guidelines for hospitals in creating a good environment, these do not usually bring together the issues for people with dual sensory impairments. Adaptations which work for hearing impaired people usually involve vision, and adaptations for visually impaired people do not usually work for hearing impaired people. Dual sensory impairment, and the multiple symptoms and conditions which rare syndromes entail mean that patients frequently have to attend hospitals but there is rarely a co-ordination for their care. Specialist commissioned clinics are likely to be a key place in which patients do receive some multi-disciplinary care, with an understanding of their conditions, but outside these, it is perhaps not surprising that Sense report that patients may not attend clinics at all, because it is too difficult, may not read clinic letters, may miss appointments even when in the hospital because they did not hear their names being called, may not receive vital information because they are embarrassed to ask for constant repeats, and may not recognise staff, (Sense 2015). Attending hospitals is rarely straightforward for anyone, but for these patients, it is even more difficult.

6 Rare syndromes and rare syndromes clinics

In this chapter we will explore the rare syndromes that are the main focus of this research project, explaining how and why they were chosen and the hospitals that they are linked to. A brief summary of these syndromes will be provided before moving on to look at what is meant by a clinic, and a brief outline of the types of clinics that participants attended.

6.1.1 Choice of participant groups and hospitals

Because we were interested in the patient experience for people with rare syndromes who have sensory impairments, we focused on six particular syndromes which frequently cause dual sensory impairments and which are all rare (fewer than one in 2,000 people have them). Amongst the considerations which led us to these groups were; that they all cause sensory impairments and frequently dual sensory impairments; that all had particular clinics or practitioners who focused on them; that several of them were ciliopathies; that there was a patient identity associated with conditions, related to, for example, the existence of a support group.

The syndromes we thus included were; Alström syndrome; Bardet-Biedl syndrome; CHARGE syndrome; Stickler Syndrome; Usher syndrome and Wolfram syndrome. We have referred to these in the following mostly as Alström, BBS, CHARGE, Stickler, Usher and Wolfram because this was mostly the terminology used by the patients.

Having chosen these syndromes, we included in our planning five hospitals which have a particular focus on these syndromes;

- Birmingham Children's Hospital which has specialist clinics for Alström, Bardet Biedl and Wolfram patients and a practitioner especially interested in CHARGE syndrome
- Queen Elizabeth Hospital Birmingham which has specialist clinics for adults with Alström, Bardet-Biedl and Wolfram

- Addenbrooke's Hospital Cambridge which has the only specialist clinic for patients with Stickler Syndrome
- Moorfields Hospital London which has a specialist service for Usher patients
- Great Ormond Street Hospital London which has practitioners especially interested in CHARGE syndrome

Please see the section 6.3 for more details about these clinics.

We later included patients who attended hospitals other than these listed above, for appointments related to these six rare syndromes, and later still patients who had other rare syndromes causing dual sensory impairments. These other syndromes were Infantile Refsum Disease, Norrie's Disease, Congenital Rubella Syndrome (CRS), and Neurofibromatosis type 2 (NF 2). There were two people with NF 2, and three individuals with the other 3 syndromes.

6.2 Brief introduction to the syndromes

This section provides a brief introduction to the six main syndromes that are the focus of this research report. Further detailed information on the syndromes can be found in the Appendix.

6.2.1 Wolfram syndrome

Wolfram syndrome was first described in 1938 and is also known as DIDMOAD - which stands for the key factors; diabetes insipidus, diabetes mellitus, optic atrophy and deafness.

Main features - It is usually noticed before age ten by patients who acquire diabetes mellitus and then optic atrophy. Diabetes insipidus and sensorineural deafness, will usually follow later. It is a degenerative condition, and people often die prematurely.

Sensory impairments – Patients with Wolfram generally have a significant visual impairment as a result of optic atrophy which means that there is damage to the optic nerve which sends messages to the brain.

Damage to the cochlea or auditory nerve causes high frequency hearing loss, usually diagnosed in the teens or 20s, and is seen in about 50% of patients.

Prevalence - Wolfram syndrome is a genetic condition passed on if both parents have a faulty gene. It affects both males and females. The overall prevalence in the UK is 1 in 770,000.

6.2.2 Alström syndrome

Alström syndrome was first identified by Carl Henry Alström in Sweden in 1959.

Main features - The whole of the body is affected in Alström syndrome with rod-cone retinal dystrophy, sensorineural hearing loss, obesity, insulin resistance, and type 2 diabetes mellitus. People with Alström may also have high blood pressure, thyroid problems, bladder difficulties, liver and lung problems, fertility problems, short stature and problems with their bones and joints. The life span of patients with Alström rarely exceeds 40 years, but early diagnosis and intervention can slow down the progression of the disease, and improve the length and quality of life for patients.

Sensory impairments - The first sign of Alström in individuals is nystagmus (uncontrolled movement of the eyes) and photophobia (sensitivity to light) in the first few months of life. As the child develops further visual loss is likely to occur. Progressive, high frequency hearing loss is often noted in the first 10 years of life; however in a minority of cases hearing loss is not noted. The hearing loss may be progressive or stable.

Prevalence - Alström syndrome is an autosomal recessive condition, which means that both parents must pass on a copy of the gene to their child, and males and females are equally likely to have Alström. Alström is a very rare condition with only 30 – 40 families in the UK affected, according to the NHS National Specialised Commissioning Team. Reports from clinical staff estimates that it may affect 100 people in the UK.

6.2.3 Stickler Syndrome

Stickler syndrome was first described by Gunnar Stickler in the USA in 1965.

Main features - Stickler syndrome is a group of conditions which affect connective tissue (collagen). Stickler is characterised by a distinctive facial appearance, cleft palate, eye problems, hearing loss, and joint problems.

Sensory impairments - There are six types of Stickler syndrome, however type 1 is the most common. The different types of Stickler syndrome relate to the genes involved. People who have Stickler are likely to be very shortsighted (high myopia), have cataracts, retinal detachment and loss of cells in the retina.

Hearing loss is found in around reported in 60% of people with Stickler syndrome. The majority of this is sensorineural hearing loss, but people also have a conductive hearing loss, or a mixed hearing loss.

Prevalence - The prevalence of Stickler syndrome has been estimated at between 1 in 7,500 to 1 in 9,000.

6.2.4 CHARGE syndrome

CHARGE syndrome was first identified in 1979. CHARGE was originally an acronym representing the diagnostic criteria (Coloboma, Heart malformation, choanal Atresia, Retardation of growth and/or development, Genital anomalies, and Ear anomalies).

Main features – As understanding of CHARGE has developed diagnostic criteria refer to problems with the eye, choanal atresia (when the nasal passages are blocked by bone or tissue), cranial nerve anomalies, and ear anomalies. As children with CHARGE get older, a number of features have been noted including curvature of the spine (scoliosis), migraine, epilepsy, cataracts, retinal detachment, delayed/arrested puberty, progressive hearing loss.

People with CHARGE syndrome frequently present with learning delay or disability, which may be related at least partly to the effects of dual

sensory impairments. However, some people with CHARGE are University graduates.

Sensory impairments - People with CHARGE often have damage to multiple sensory systems, including smell and tactile senses as well as visual and hearing difficulties. They frequently have difficulty with balance and controlling their bodies in space.

Ear abnormalities not only affect hearing but also balance and, accompanied by low muscle tone, as well as a visual impairment, may mean that people with CHARGE have difficulty walking,

Vision problems are generally related to coloboma, where there is a gap in the structure of the eye.

Prevalence – CHARGE is a sporadic autosomal dominant condition, although there are a few incidences of parents who have mild forms of CHARGE passing it on to their children. The incidence of CHARGE in the UK is between 1 in 10,000 and 1 in 15,000 births.

6.2.5 Usher syndrome

Usher syndrome was first described by Dr von Graefe in Berlin in 1858 with the name coming from Dr Charles Usher in the UK in 1914.

Main features and sensory impairments - The predominant features of Usher are sensorineural deafness and progressive vision loss due to Retinitis Pigmentosa (RP). Initially peripheral vision loss occurs which is described as 'tunnel vision'. People with Usher also have particular difficulties in seeing at night time (night blindness) or poor lighting conditions.

Three 'types' of Usher are usually described (and within these subtypes exist) – Usher 1, Usher 2 and Usher 3. Typically the three types are used to differentiate the severity of deafness (Usher 1 connoting severe-profound deafness, Usher 2 moderate-severe deafness) the age of onset of vision difficulty (Usher 1 in early teenage years, Usher 2 in later teenage and Usher 3 in middle age). Usher 3 is then described as including progressive deafness (not so for Usher 1 or 2) and Usher 1 and sometimes Usher 3 include balance difficulties.

Prevalence - The exact prevalence of Usher syndrome is unclear however figures from Sense suggest it might be as high as 15 per 100,000.

6.2.6 Bardet-Biedl syndrome

Bardet-Biedl syndrome is named after George Bardet and Arthur Biedl who in the 1920s identified individuals who shared retinal degeneration, post-axial polydactyly (extra fingers and/or toes on the outside of the foot or hand) and obesity.

Main features – Obesity is a very common feature of the syndrome and the development of type 2 diabetes mellitus is also prevalent. Developmental delay is seen in a number of patients – but not all. Kidney problems and genital abnormalities are also a feature

Sensory impairments – Conductive hearing loss is seen in around 20% of patients. Within the first decade of life vision problems begin to occur, with a gradual loss of vision as time goes on. This is due to rod-cone dystrophy, where the rod cells in the eye, followed by the cone cells are lost.

Prevalence - Bardet-Biedl syndrome is traditionally thought to be autosomal recessive although there may be some variations. In the UK the prevalence is estimated as 1 in 160,000 people.

The expression of all of these syndromes differs from person to person, and even within the same family there were differences in the ways in which the syndromes present, as Muna, Jonathan, and Angie explain:

It affects them all different. Deema it's her eyes. Her vision. She's lost it all now nearly. The peripheral and she can't see at all in the dark or the bad light. But she's learning at school how to find the tags on her clothes and things so that she can put them all on the right way round. She's got a fatty liver. Aalia she's got the eyes, but she's also got Perthes disease which is in her hip, so one leg is shorter than the other, and she's really unsteady, and she's got the brace thing on her leg. They've got diabetes, ...Lilas she was born without a vagina. When she was born, she was in ICU for 18 days

because of all the urine was around her and she couldn't breathe. So now she has a catheter into her belly. She wears nappies at night. She's had 3 operations. She's got one more to go when she's a bit older. Her eyes aren't as bad as the other two. (Muna, mother of Deema, 14, Aalia, 12, Lilas, 9, BBS).

In CHARGE syndrome someone doesn't always have the same symptoms as someone else with CHARGE. (Jonathan, 12, CHARGE).

The thing I find difficult about the advice for Stickler is that it is such a spectrum and some people are driving cars and some are really disabled through it. (Angie, 43, Stickler).

6.3 What is a clinic?

The participants within this project attended a range of different clinics which can be grouped into the following three categories:

- Specialised holistic services
- Condition specific clinics
- General clinics

Participants also then attended therapy appointments such as physiotherapy, hydrotherapy, and speech and language therapy.

Please note that these terms are used by us, in this report, to provide clarity and organisation in exploring the experiences of people with sensory impairments as they access clinics in relation to their rare syndrome, nevertheless they broadly represent the structure and format of the various clinics attended by participants. These are not terms used in the NHS to designate these types of clinics.

As well as the hospital's name, participants also used the doctor's name (e.g. Mr Z's clinic, or "when you go and see Bob Smith"), the organ under investigation (e.g. "your eyes", "my kidneys"), the hospital's location "when I go to Birmingham", or the person's job title (e.g. "the dietician", "the psychologist"). Participants were generally not aware of

the subtle differences in the way that their care was being provided e.g. in terms of funding, or what might be available at other hospitals.

6.3.1 Specialised holistic services

Specialised holistic services are those clinics which are provided on a yearly basis and bring together a range of specialisms (e.g. cardiology, endocrinology, ophthalmology, psychology, dietetics) all in one place. It is often described as a 'carousel service' by clinicians as a patient will attend as many consultations as needed on this day. (The adult Alström service at the QE hospital in Birmingham runs over 2 days).

The patient will continue to see their local doctors and attend various other check-ups and appointments throughout the year. One of the lead consultants at BCH described this service as a '30, 000 mile MOT' - it is an annual review to get a broad picture of the health and needs of the patient, and is not meant to replace local services.

These clinics are in one location in the country and patients will generally have to travel a considerable distance to attend the service. Because of the low incidence of the syndromes and the complexity of need all patients with the syndrome are invited to attend these clinics.

In relation to this study the following clinics can be described as specialised holistic services:

- Adult Alström service – QE Birmingham
- Adult BBS service – QE Birmingham
- Adult Wolfram service – QE Birmingham
- Children's Alström service – BCH
- Children's BBS service – BCH
- Children's Wolfram service – BCH

There is also an adult BBS service running out of Guy's Hospital London, and a children's BBS service at GOSH.

6.3.2 Condition specific clinics

A second type of clinic that participants attend is that in which a consultant and his/her team will focus on a particular condition (e.g. Usher, Stickler, CHARGE), and through the lens of their speciality (e.g. ophthalmology or endocrinology) will provide clinical expertise for the patient. However the other aspects that make up that syndrome are not addressed at that particular time or by that doctor.

Often people will have to travel long distances to get to these clinics and a special case for funding to attend is often required.

In relation to this study the following clinics can be described as condition specific clinics:

- Usher – Moorfields
- Stickler - Addenbrooke's
- CHARGE - BCH

There is some evidence (from participants) of rheumatology and audiology being provided at the same time (on the same, previous or following day) for patients with Stickler at Addenbrooke's hospital, but it is not co-ordinated in the same way as the holistic services and appears to be rather *ad hoc*.

6.3.3 General clinics

‘General clinics’ is the term used to describe clinics that are neither specialised holistic services nor condition specific clinics. They are still likely to be highly specialised (although perhaps not nationally leading) and are usually based at major teaching hospitals closest to the participant.

Depending on the needs of the participants and local provision, participants may attend general clinics at more than one hospital.

Participants are very likely to attend general clinics if they also attend a specialised holistic service. Participants are also likely to attend general clinics for the other aspects of their syndrome if they go to condition specific clinics, but not for the speciality which is the focus at the

condition specific clinic .i.e. if a person with Usher sees an ophthalmologist at Moorfields they are unlikely to go to a general ophthalmology clinic, however they would probably attend a general audiology clinic.

Participants sometimes also attend clinics for conditions unrelated to their rare syndrome/disease, or had attended them in the past. Where relevant, in order to obtain a full understanding of the experiences of the participants, this information was also recorded.

In relation to this study we specifically looked at general clinics at Great Ormond Street hospital in relation to CHARGE syndrome, and participants also gave their experiences of general clinics as well as those of condition specific or specialised holistic services.

7 Methodology and ethics

7.1 Methodology

This section outlines the methodologies and methods undertaken for the study to gather as much data as possible about the experiences of people with rare syndromes and sensory impairments attending specialist clinics.

The main aim of the study was to examine the experiences of people attending specialist clinics. This was intended to go explicitly beyond 'measures' to find out what being a patient is like. This led to a qualitative paradigm, designed to find out in rich detail what people with dual sensory impairments themselves wanted to say about their experiences. Luxford and Sutton (2014) had suggested that these voices might have a more powerful impact on medical practitioners than lists of numbers or figures. These individual stories present the feelings and perceptions of the patients, and this individuality was part of what it was intended to reflect; the very nature of rare syndromes and sensory impairments being that no two patient experiences are the same, and the needs of each person are different. Because of the nature of the participants (some very young, some older, all with sensory impairments and frequently with access needs for communication) and the nature of the clinics visited; some diagnostic, some an annual overview, some short (half an hour's review) and some long (spread over two days) a very flexible approach was needed. The methodology and the methods were therefore chosen to allow for great flexibility, attention to individuals and individual differences and to give people the opportunity to say the things they wanted to.

Gathering individual perspectives and being informed directly by the users of the clinics was at the centre of this study. It was not intended that we would compare hospitals to each other, as people respond in different ways to the same hospital and have different experiences. Instead, the study draws out examples of good practice, where patients have left hospitals with improved understanding of their conditions and have felt that the hospital has been attentive to their needs, but also

demonstrates what difficulties they have experienced, so that feedback can help hospitals to develop their services.

The approaches through the frame of individuality therefore led to mixed methods within an ethnographic approach (as suggested by LaVela and Gallan 2014). While they included some elements of case study, elements of the 15 Steps approach and shadowing were also used (Thomas and Clarke 2015 and LaVela and Gallan 2014), in terms of environmental audits and observations. These allowed for real time, real patient experience, not simply drawing on what was remembered, in which many details might be missing. Elements of what was not present may be more likely to be represented in more objective audit than in patient recollection. This draws on a multi-dimensional way of gathering information about individual experience, such as described by Thomas (2011) in relation to case studies, and allowed the triangulation of data. At the heart of this study and in line with the funder's intentions, are the patients' stories, but the data from accompanying patients in real time and in auditing clinic environments strengthens and enriches their voices.

7.2 Methods

7.2.1 Interview

The first part of the study involved a semi-structured interview. Interviews were recorded with explicit participant permission. Interviews provide the participants with the opportunity to talk about themselves with someone who was attentive, sensitive, non-judgemental and used prompts and checks to ensure that they had understood (Denscombe, 2003). It also enabled good communication techniques to be used (appropriate level of speech, use of BSL, and so on) . This interview was designed to give people the opportunity to talk about their own experiences and the researcher's role was to frame this within their lived experience as a person with sensory impairments and with a rare syndrome. For participants, these two issues were so infused with their personality that they did not always think of their lives in this way. For this reason, it was important to ensure that these issues were part of the discussion. Therefore the semi-structured questions in the interview

focused on the three issues at the centre of difficulty for most people with dual sensory impairments – communication, information and orientation and mobility. The areas investigated included travel and mobility to and within the hospital, the information provided before and after the clinics, and communication from receptionists, clinical staff and others.

For example, questions were included (at the discretion of the interviewer, given their individual circumstances) about whether people had to travel between rooms during the clinic, whether clinic rooms were noisy, whether a guide dog could be accommodated, and what lighting and signage were like.

Questions were asked about accessible formats for test results, opportunities to examine equipment by touch before it is used, information about delays, and the process of making appointments. The interview included asking about how staff communicated, how much they understood about the need for individual communication methods, including extra time, and whether they understood and used appropriate methods for instance, for guiding, and whether staff spoke to the patient or to someone supporting them.

As well as all this, other important issues were raised, such as who supported patients in clinic, how those accompanying were treated, the understanding of the role of interpreters, and whether the clinic acted as a point of contact with others with the same condition. Additionally some demographic data and information about individual syndromes was collected.

7.2.2 Environmental audits

As a context and background to the data collected from interviews we carried out environmental audits at the five hospitals identified in section 6.3 above. Our environmental audit was intended to examine the hospitals in relation to key issues for people with dual sensory impairments, that is, related to vision – e.g. glare, lighting, signage; to noise; to mobility – e.g. ability to trail, use a cane, accommodate a guide dog; and to the specific areas which patients needed to use, such as corridors, waiting areas, toilets and cafés, and clinic rooms.

Environmental audits for visual impairment have been used in education (e.g. Naish and Clunies Ross 2004) and likewise for noise (NDCS 2008) to help examine an environment and recommend improvements.

Although environmental audits of hospitals do exist (for example, Patient Led Assessments of the Care Environment, NHS 2013) they do not necessarily include all aspects of the environment which are relevant to people with dual sensory impairments. We developed and used an environmental audit which included parking and access to the hospital, routes through the hospital, signage, toilet areas, reception areas, the use of alternatives/supports to vision/hearing (such as loop systems, braille notices, staff preparedness to communicate,) refreshment areas. We gathered the data for the audits in some different ways; some of this was through invitations by hospital staff to attend specialist clinic days, some was through accompanying participants when they attended hospitals, and some was through walking through and round hospitals to collect this data specifically.

Eight environmental audits of clinics were conducted in 5 different hospitals. Five of these environmental audits took place at clinics that were specialised holistic services, 3 were condition specific clinics, and 1 at a general clinic. (See section 6.3 for a definition of services).

7.2.3 Accompanying participants on visits

In order to gain as real a sense as possible of the patient experience, of the individual difficulties faced by some participants during their hospital visits, and of their needs as sensory impaired people within the hospital environment, and how these were addressed, we accompanied some patients on visits to clinics (as in LaVela and Gallan 2014). During these visits we collected data for the environmental audits, for additional interview material (when patients talked) and we also completed an observation sheet. This gave us some insight into how long procedures took, what communication methods staff used, how aware staff were of the needs of people with sensory impairments, the time given to communicate, including reading papers, and more. These visits were rich in terms of data which was recorded as interview data, environmental audit and some as observation. We accompanied five patients on their clinic visits (two children and three adults), all of whom were dual sensory impaired. They took place at four different hospitals.

7.2.4 Analysis

To analyse interview data, the interviews were transcribed, using strict ethical protocols, pseudonyms, and leaving out information, as far as possible, which could compromise individuals' identities. It is recognised however that given the very rare nature of some of the syndromes, some participants may be recognised by people who know them well.

The transcriptions were then added to a software driven analysis tool *NVivo* which enabled the research team to draw out key ideas and themes, both those they were hoping the questions would identify, such as mobility, communication, and information, but also others which arose from participants during the analysis, such as connectivity and individuality. The tool allows for continual comparison and thematic analysis.

Environmental audit data was tabulated under the headings of the audit and then presented in such a way that individual hospitals were not linked to specific features. These were then linked to the thematic analysis. Data from observation during the individual clinic journey was also linked to provide specific examples in relation to the themed analysis. We have presented the data in the order of a clinic visit; appointment, at the hospital, at the clinic, consultation, beyond the clinic, and additional points.

Data pertaining to the environmental audit is identified as hospital 1), 2), 3), 4, and 5) when relevant overall to a hospital, and is labelled clinic (i), (ii), (iii), (iv), (v), (vi), (vii) and (viii) when documenting individual clinics. At clinics (i) and (iv) more than one space was used so these are identified as a), b), and c). The accompanied clinic visits are referred to as a), b), c), d) and e).

7.3 Ethics and participants

The aim was to include people from each of the syndrome groups and also who attended each of the possible hospitals. Because the study intended to include work on NHS premises NREC ethical approval was obtained in June 2014.

In order to accompany patients on hospital visits and to carry out environmental audits and specialist clinic visits we then obtained ethical approval from each of the hospitals we visited.

We intended to include as wide a range of people as possible, older and younger, and those with and without learning disabilities. Except for a few parents of very young children or children/adults where we only met family (for whom no interaction with the sensory impaired person was included), all who were consenting or assenting had specific information needs related to their sensory impairments.

Although we developed materials to gain consent relating to adults who were not of themselves fully competent to consent for themselves and for consultees on their behalf, all our adult participants in the end were fully competent. Some required information in other formats (e.g. braille, large print, BSL). All the materials for recruitment and study were therefore potentially available in a range of formats. Where BSL interpretation was needed this was arranged with the participant.

For children, consent was sought from parents/guardians but assent was also sought from children themselves where they could be asked. We had information sheets for children (from about 5 years old) and for those children who were 12 years or more we also asked more formal consent.

7.3.1 Consent

No-one was or would have been excluded because of literacy and communication difficulties. We had developed materials which outlined the study and which asked for agreement for people who used symbols or simple text, as well as signs, and we were ready to adapt these for individuals (NIHCE 2012, Robert et al. 2009). The interviewers were trained in a protocol for checking whether participants had the ability, under the Mental Capacity Act, to consent to this study. If they were not able to, we had in place a procedure for consultees to give consent on their behalf. All our arrangements were approved by the NRES committee because we believed it was very important that wherever possible patients were asked their opinion, however old they were, and whatever difficulties they had, if they could remember something about their appointments and explain it.

7.3.2 Recruitment of participants

In order to recruit participants a number of key contacts were used. This included,

- Syndrome specific groups; (such as Wolfram Syndrome UK support group and CHARGE family support group). These groups were approached to support the study and some advertised on their websites, some invited us to attend specialist events or conferences to discuss our project and some also recruited through personal contacts.
- Specialist staff; teachers of hearing impaired, visually impaired and deafblind children were asked to pass on information to appropriate families.
- Specialist provision; special schools, colleges, and other providers were asked to pass on information about the study to appropriate individuals or families
- Publications, internet and social media; Sense published information in their members' journal and in their magazine, and information about the study was also placed on social media, the University of Birmingham Deafblindness team's Facebook page and on the research page, as well as internet and social media sources for individual syndrome groups, as above.

7.3.3 Participant numbers and demographics

52 people were interviewed and told us of their experiences of rare syndromes, clinics, and sensory impairments; of these, 42 were people who have a rare syndrome, and the remaining 10 were from parents/guardians of people with a rare syndromes speaking about the experiences of clinics for the people they care for and in their own role as a parent or carer.

For purposes of clarification in the report we use 'patient' to describe the person with the syndrome, and 'participant' to describe the person who is taking part in the research, who may or may not have the syndrome.

In two of the interviews parents also contributed to the interview where their child was the main participant. In a further four interviews a partner/ close relative also contributed to the participant's experiences. In four of the interviews with people with Stickler the participants were speaking not only as a person with Stickler, but also as parent/carer of a person with Stickler.

We interviewed two siblings with Wolfram, two siblings with Stickler, a mother with Stickler and her two children who have Stickler, two siblings with BBS, another two siblings with BBS as well as their mother who spoke about a third sibling's experiences in addition to her viewpoint as a mother of 3 children with BBS. These interviews happened simultaneously. We also spoke separately to a brother and sister who both have Stickler.

The patients were aged from 14 months to 83 years old.

20 of the patients were male, and 32 were female.

Table 1: Participant numbers, age, and gender according to syndrome type

SYNDROME	No. of patients	Age of patients (years)	No. of female/ no. of male
CHARGE	9	14 months – 18	4 Female 5 Male
Usher	11	34 - 83	7 Female 4 Male
Stickler	13	6 – 48	11 Female 2 Male
Alström	1	49	1 Male
BBS	8	8 - 60	5 Female 3 Male
Wolfram	5	21 - 55	3 Female

			2 Male
Other	5	7 - 45	2 Female 2 Male

Patients were from a range of different ethnic backgrounds: 36 described themselves as White British, 4 White British (English), 3 British Pakistani, 2 Black British, 2 Pakistani, 1 White British (Welsh) and 1 White Other. Data was not obtained for 2 patients.

Participants had varied and interesting lives, having a rare syndrome and attending clinics was not the main focus of their life, although some participants were more readily able to position themselves as a patient than others. As well as being patients, participants spoke of being sons and daughters, pupils, musicians, cooks, friends, husbands and wives, mothers, campaigners, sportsmen, artists, and many more identifiers. We would not wish to forget this rounded perspective in our descriptions of the small element of their lives that was as “patients”.

8 Analysis of patient experiences of clinical settings

The experiences of participants were explored to highlight what makes the delivery of services both efficient and effective for people with sensory impairments, as well as the factors that might inhibit this. Data is drawn from the 52 interviews with participants, the eight environmental audits covering five different hospitals, and 5 accompanied clinic visits.

Five key moments in the experiences of participants attending clinics were explored – the appointment, at the hospital, in the clinic, the consultation, and after the clinic - these will be discussed in this chapter along with a further section on other issues which emerged from patients' discussion of their experiences.

8.1 Appointment

8.1.1 Getting a referral

The process of getting a referral to a particular clinic whether it be a specialised holistic service, a condition specific clinic, or a general outpatient clinic was different for different people. For those attending specialised holistic services, getting a referral seemed straightforward and participants usually attended the clinics that were offered to them. Greater difficulties were experienced by participants who wanted a referral to a condition specific clinic, often this was related to funding, as Harriet, a woman with Stickler and also a mother of two children with Stickler explained:

So I wanted to go as I read about cryotherapy on the internet and (major local hospital) didn't want us to go as it was an out of county referral and it was very expensive and they didn't want to do [it] and they didn't think cryotherapy would be successful. So he didn't want to do it so I had to throw my toys out of the pram to get a referral. I said it wasn't his choice – it was up to me to make the choice. And if he could refer me so at least I could find out more rather than not

knowing. In the end he did refer me to go. (Harriet, 38, Stickler, mother of Chloe, 11 and Ollie, 6 Stickler).

Louise (41, Usher) also had similar difficulties, getting referred to a specialist Usher clinic due to the fact that there was not a clear plan of how referrals in her local area worked, and a mix up with paperwork at the CCG (Clinical Commissioning Group) lead her to describe her experience thus, “it was complicated. It is very convoluted really.” Ultimately however on having received her appointment she explained that she was “delighted”. It is important to note that for Louise, up until this point, she had not been ready to deal with the emotions associated with deteriorating vision and a dual sensory impairment, and it was only when she began to engage with other people with Usher on Facebook, and in real life, and began to hear their experiences, that she herself chose to request to attend a condition specific clinic:

...in the past year I have been more proactive and I have asked to see [consultant] at Moorfields... It seems that nearly everybody goes to Moorfields. I thought I wanted to go to Moorfields. I thought I am going if everybody else is going. (Louise, 41, Usher).

No matter what clinics and services are available, if patients are not ready for them they are unlikely to attend or if they do attend, they are unlikely to find them useful.

As seen with Harriet and Louise, a number of participants had carried out their own research on their condition or treatment options and had requested to see a particular specialist or attend a specific clinic as Chris and Sally explain:

We specifically asked to be referred to them because we heard that they had such a good reputation around cochlear implants. (Chris, 31, Norrie’s Disease).

...I had asked about some treatment that I had seen on the CHARGE Facebook group about stabilising retinas through laser treatment, so the [local] ophthalmologist wrote to Great Ormond Street and said would you see this family. So we did and first saw

Great Ormond Street in June of last year. (Sally, mother of Poppy, 5, CHARGE).

As will be discussed later in this report, specialist syndrome specific conferences were very useful to patients and families to learn more about the syndromes. It was at a CHARGE conference that Leo's mother Sophie learnt about the work of a consultant running a condition specific clinic from a conversation with the consultant on the day. Sophie immediately asked for a referral which went ahead but she was frustrated that this specialist working at a hospital relatively close to her had never been mentioned to her previously:

...I can't believe that he's just in [city], and he's been doing CHARGE for 20 years and no-one has ever mentioned him. I then was pushing to get [consultant] because he's got all the knowledge in the world about it. He goes to the States and everywhere. He's been great. He's really good at explaining things in a way you'll understand rather than too technical. He's really been great [consultant] has. (Sophie, mother of Leo, 17, CHARGE).

Whilst Sophie and Leo were very appreciative of this referral, sisters Alexa (27) and Roxy (29) who both have Stickler (along with their father and Roxy's children) felt trapped by a referral to a condition specific clinic. This clinic was far from their home town, making it very difficult to attend, but they felt compelled to attend as they needed to be seen somewhere and their local hospital would no longer see them. Alexa explains:

I went to [local eye hospital] and once everything is referred to [condition specific clinic], they refuse to see you at [local eye hospital]. It is all a bit of pass the buck situation which is tricky, you know when you really want to be able to go somewhere local because there is no real reason why they can't check your eyes in [local hospital]. Just regular check ups to look in your eyes. There is no need to just have it once a year all the way in Cambridge. I don't think there is anyway. (Alexa, 27, Stickler)

As Alexa and Roxy's circumstances demonstrate, and will be discussed further below, once funding has been arranged and participants get a

referral, and a patient gets an appointment, a lot of things need to come together in order for that person to be able to attend their appointment.

8.1.2 Making an appointment

There was no standard procedure for making appointments; sometimes participants would receive an appointment in the post, either just after their last appointment or a month or so before the appointment was due, at other times they would make an appointment at the end of their consultation. Often different clinics in the same hospital would use different procedures.

Patients, or their families, usually knew which times of year their appointments would be and this helped to provide structure to the year as Sandra, mother of Max commented:

We tend to usually get a letter through the post. Sometimes I've had it where you have to ring up and book at a convenient time. I know which ones are afternoon or morning clinics now. March and November are my two six monthers so I know that they are coming up. So I had quite a few recently but they are tailing off now.
(Sandra, mother of Max, 11, CHARGE).

Nina (46, Usher) had specifically chosen to have make her appointment in May due to the lighter nights, and Jeff (42) who also had Usher was considering asking whether it would be possible to move his appointment from January to a 'summer month'.

For some participants, on receiving an appointment through the post there was a sense of frustration and lack of understanding of their needs in the times and dates that they were given. Dave (43, Stickler) was not only frustrated by the lack of organisation and connected thinking when organising appointments, but believed that better organisation would lead to a more efficient and effective clinic for all parties:

They gave me an appointment for 8.30 am start... from here! Which shows the complete lack of consideration of the person booking the appointment... It's just the awareness of administration teams taking a moment to look at the address and actually say well people living in [local area] would find it easier to get in to the appointments in the morning, early morning starts. It may benefit them more so offer those times to them and give a bit more time for other people to come in from further afield. That kind of logistical thinking is still weak. (Dave, 43, Stickler).

Harriet (38, Stickler) dealt with these frustrations by “getting involved” and “not just accept[ing] those appointments”. As will be discussed later in this report, for families attending clinics it is often useful to have family appointments, where everyone can be seen at once. For Harriet two of the clinics at different hospitals that she attended with her children provided family appointments but at the third hospital this did not happen, “they tried to give me an 11 o'clock for him and a 2 o'clock for her so you have to hang around 2 and a half hours in between which is ridiculous!”

Jean (67, Usher) however suggests that there is no need for such frustrations if a patient is able to make their appointment themselves and with plenty of advance warning:

I had the control not them. At [current hospital] they have the control and not me. And I much prefer the [previous hospital] way, because if I knew a date a year ahead I could work out when not to go on holiday etc. So it was much better. (Jean, 67, Usher).

However as Wendy suggests even when things have been booked up far in advance there can still be frustrations:

I'm quite an organised person so we will book things in advance, like we are going away at Easter – I have booked it around his appointments. And then I get really frustrated when they ring up and say ‘ooh can you change this appointment’. Well I can, but you know what I have just based my holiday around this. And this does

happen quite a lot. You get ‘oh it’s such and such from [hospital] and my heart sinks’. (Wendy, mother of Luke 11, CHARGE).

Particularly for those patients that are attending specialised holistic services or condition specific clinics, which continue over many hours and are often a long distance from the patients’ home, advance notice of appointments was particularly important, as Kevin, uncle of Isla explains:

I think we are going to the clinic in August. That’s what we’ve got to get in touch with [them] about because we need to book the hotel and we can’t do that on a spur of the moment thing. Because you have to book. (Kevin, guardian of Isla, 8, BBS).

It is also useful to note that Kevin uses the expression “I *think* we are going to the clinic in August” as often participants would *think* that they had a clinic appointment coming up but it would not always occur and patients would be left without any clinical support as the examples from Holly and Jen demonstrate:

For my hearing, I thought I would expect to have my hearing test every year. It is now past 2 years and I am still waiting. I don’t know what the situation. With my eye sight, it is just a matter of waiting till I get a letter. You know, they said 12-18 months but the waiting list is getting longer and longer. (Holly, 49, Usher)

For her eyes she just goes to SpecSavers. When she was little she always had to see an orthoptist. It just seemed to fizzle off. They don’t seem to send you another one. I don’t know if it’s me, or if it’s just them not sending me another appointment. (Jen, mother of Megan, 18, CHARGE).

A small number of participants also experienced difficulties when for some reason they had to cancel an appointment. For example, when Jack (27, Wolfram) cancelled his ophthalmology appointment at his local hospital he was not provided with an alternative date, although they “they said they would send me a new one, but I never heard back from them.” Similarly Gordon (34, Usher), although he had previously been going once a year (due to a retinal tear) was advised to be seen every one or two months. Unfortunately due to his work commitments as a teacher he was twice unable to make the appointments and, despite

letting the hospital know, he was discharged from the hospital and had to get a re-referral via his GP:

...it wasn't as though I was ignoring them, I did ring up and say I'm sorry I can't make this day. They didn't come back to me at all, but after the last one I just didn't get a new appointment. And I phoned them up and they said oh no because you haven't attended you need to go through the GP again. You need to get the GP to refer you on. So I was saying, you're telling me that I've got a tear at the back of the eye, this is really urgent, I need to be coming back and you're telling me that I can't go back. I've got to go through the GP. I went through the GP again. I went about 2 months ago, and I'm still waiting for an appointment. (Gordon, 34, Usher).

For Gordon this process was extremely stressful and worrying and appeared to him very unfair, however for Basil having the opportunity to change the appointment a maximum of three times was viewed as quite reasonable:

They are quite fair. They give you 3 goes. You can change it once, you can change it twice, no more, after that you are off our list. You can only cancel twice. You have to go back to the GP and completely re-register. It's fair enough. (Basil, 57, Usher).

A small number of participants also spoke of specifically choosing not to attend certain appointments in order to make their life easier, often balancing the effort of going to a clinic against what the doctor would be able to provide for them. For example Jonathan's (12, CHARGE) parents, although they kept up with certain appointments when he was young, decided not to follow up with genetic testing appointments as they were not essential in keeping Jonathan healthy, and furthermore a reduction in the number of appointments would mean less stress for the whole family.

Vera (83, Usher), although she had been to a condition specific clinic in the past, had now decided against continuing to attend as she had got what she wanted from it (certification of visual impairment and a genetic diagnosis of Usher) and as an older person a long journey with an overnight stay, was on balance not worth it:

I felt that I'd had enough of it. It is too far to go. It takes a lot out of me. And I know what's happening. They've done the blood test. I know where I stand. That's enough. (Vera, 83, Usher).

8.1.3 Accessing appointment information

Having been given an appointment participants spoke about the struggles of accessing the information pertinent to the appointment e.g. date, time, and location. Being able to access the information on an appointment letter is crucial to being able to attend, as well as for promoting independence, and when this does not happen it can be very frustrating as the following examples from Doug, Angie and Chris demonstrate:

I have asked for email but they don't tend to work that way. And my mum is not good at reading things now. It's a bit awkward. She manages to read it... or my brother comes once a week to read things. It's just a bit awkward, I would just prefer to have it by email if it was possible, but they don't seem to do that for some reason. (Doug, 60, BBS).

They send me appointment letters how they normally send it. They don't email. They send it in these awful things, they're not even envelopes you have to rip the sides and open them. I find that inaccessible in itself. I have to rely on support to access appointment letters. Once I am told the information I put it on my diary sheet on my computer which means I can access it but not the actual way that they send it to you. (Angie, 43, Stickler).

Generally yes, I would prefer to have appointments by email or a format that I can read that would be more appropriate. If I lived on my own it might have been more difficult. It's sort of the one thing I would say, the only sort of minor criticism. (Chris, 31, Norrie's Disease).

It is interesting to note that Doug, Angie, and Chris were speaking of their experiences at the three different 'types' of clinic as mentioned previously (Doug – specialised holistic service, Angie – condition specific clinic, and Chris – general outpatient clinic).

What is more frustrating, as in the case of Doug and other participants, is that they had specifically requested and asked for alternative forms of communication and it did not happen. In another case, both Jack (27, Wolfram) and Dave (43, Stickler) had begun receiving appointments by email but at some point that tailed off and they no longer received appointments in their preferred format. Nevertheless some participants did receive information about their appointment in their preferred format, for example Rob (27, BBS) received information in large print and also as a back-up via email. Due to the fact that Simeon (46, Usher) received his appointment information in an accessible format this boosted his confidence in attending this particular clinic as he believed that this demonstrated their interest in him as an individual:

Yeah because they made the effort to make everything 20+ letters. That's why I am always confident going to [hospital]. I don't really have a fault going there. (Simeon, 46, Usher).

This is also exemplified in Louise's (41, Usher) experience when she was independently able to confirm her attendance at her appointment:

...they sent me a letter, but with a mobile number to say text to confirm your appointment attendance. That is the only time that has every happened so I was given a reference number and I just had to quote the reference number and you know day my appointment was, and yes confirming my attendance, and they texted me back to say 'just to let you know we have received your confirmation.' But I did that without anybody helping me! [husband] didn't have to ring them up, and I was literally gobsmacked. I thought I can sit here and text them and say "yes I am coming"! (Louise, 41, Usher).

As well as information regarding the time, date and location of the appointment Nina suggested that it would be useful to have some more details regarding the appointment, for example, the dilation drops, the retinal photography, that it does involve periods of waiting around:

I think if they sent out a letter in the future they should explain that you will see different people, there will likely be a nurse to do this – this kind of thing – just so that they know that there will be waiting around and a bit to and fro. Cos it is a bit confusing and sometimes you are asked to wait in different areas, so that makes it confusing

because the guy will say well you can wait out there, or you can wait here if you want and you kind of get a bit worried because is the other guy going to come and find me then? So that can be a bit on edge... but I am kind of used to it and they will probably come and find me and my husband is with me, but if someone was on their own it would actually be awful. (Nina, 46, Usher).

This was echoed by other participants, also speaking about ophthalmology appointments like Nina. For example, Sally (mother of Poppy, 5, CHARGE) commented that it was only after attending a couple of times that she was beginning to understand the difference between the orthoptist, the optometrist, and the ophthalmologist, and Basil (57, Usher), Gordon (34, Usher) and Dave (43, Stickler) made the point that the patient does not always see the consultant and this can be confusing at first for patients. For instance Jeff, in response to the question “what other information would you like?” replied:

Only really [with regard to] seeing the doctor that we are supposed to be seeing, not someone else, because it gets confusing when I am expecting to see [consultant] and then I’m seeing other doctors. [Consultant] sort of comes in towards the end of it. And I thought that he was the main doctor that I was supposed to be seeing. (Jeff, 42, Usher).

8.1.4 Managing appointments

One of the biggest difficulties for participants was managing the sheer number of appointments as well as juggling the various factors such as childcare, time off work and school, and travel logistics that are necessary in order that a person can attend an appointment. This was particularly so for patients with CHARGE, or those attending general clinics alongside specialised holistic services and less likely for adults with Usher. For example Nina (46, Usher) attended an ophthalmology appointment once a year but had not been to the audiologist for 6 years, similarly Jean (67, Usher) saw an ophthalmologist once a year but it had been two years since she had seen an audiologist. Basil (57, Usher) had an ophthalmology appointment every 18 months, and a yearly audiology appointment. However Paul (49, Alström) as well as attending a yearly specialised holistic service where he saw an audiologist, psychologist,

cardiologist, lung function specialist, and kidney specialist also attended a local hospital on average 12 times a year for hormone injections, feet checks and blood pressure monitoring. Helena (mother of Rosie, 4, CHARGE) listed 15 specialists that her daughter saw across three different hospitals, and a local children's centre and Josh (13, CHARGE) attended 8 specialisms across 4 hospitals and one children's centre. For patients that have more than one medical condition, for example as well as having BBS, Isla has epilepsy, an even greater number of appointments will need to be managed. For example, Kevin (guardian of Isla) showed the researcher a list of 22 names of medical professionals who are working with Isla.

Juggling the volume of appointments was tricky, not just medical appointments but all those that go together in managing a complex condition or caring for a child with significant health and medical needs. Wendy for example is quite candid that it is only down to her own personal circumstances that she is able to ensure that her son Luke (11, CHARGE) attends the necessary appointments, "it's only because I am home fulltime that we juggle all the balls with all his appointments."

Kirsty for example explains how her son Finlay (14 months, CHARGE) had to miss a kidney scan due to a prior appointment with a Social Worker:

You have to try and mix and match them and make sure none of them clash on the same day... And if they do clash I have to make sure he goes to the most important ones and then try to re-book everything else in between. (Kirsty, mother of Finlay, 14 months, CHARGE).

In such instances it is parents who make a judgment as to which is the most important appointment to attend.

The pressure of having two appointments on the same day can be exacerbated by the fact that clinics often overrun, however with prior negotiation with the teams involved Diane was able to ensure that her son was able to attend both appointments on the same day:

...no if his appointment card says 10 o'clock, it's got to be 10 o'clock, you've got to see him on time, because at half past ten,

we've got to be the other side of (city), and parked, and in the hospital, because the guy's down from [London] to run his cardio clinic, and he's only there once a month (Diane, mother of Dean, 29, CRS).

For Wendy (mother of Luke, 11, CHARGE) combining the appointments all on one day has been useful as she suggested, "The most that I've ever been able to do is get 3 appointments on the same day, and it was a long day, but it was one day rather than 3." The advantage of this was that her son has had to miss fewer days of school, which is not only important for his education but his routine and structure in daily life. Wendy mentions that it would be a lot easier if there was a combined approach to appointments:

I think it would be easier if it was all in one place, all in one day, because I find it incredibly frustrating when for instance, this has happened quite a lot recently, in terms of appointments one day after another, for example I'll be in on a Tuesday to see his gastroenterology team and then in on a Wednesday to see his endocrinology team. And that is really frustrating! Because irrespective of the energy of getting up there, it's the loss of structure for him which is really disruptive, and having the opportunity to have things co-ordinated would make life so much easier. (Wendy, mother of Luke, 11, CHARGE).

Participants who were mothers of children with CHARGE also recognised that for parents of children with complex needs and seeing numerous clinicians, (medical professionals, education specialists), having some visit a home setting or at school was particularly useful, especially in the younger years:

A lot of the developmental people used to come to our house instead us going to them for appointments. And that made a big difference at the time. Like the [MSI advisory teacher], the physio, the OT, the health visitor. People who could come to our house did. And that was really helpful when we were doing 3 or 4 appointments a week. (Helena, mother of Rosie, 4, CHARGE).

If you had to average it out over the last year [we] have an appointment every three weeks or something, but that's so much

reduced from when he is very little. Not just hospital appointments but seeing the physio, speech and language therapists, teacher of the deaf, there was something every week. Usually something every week. But because a lot of the physio and things like that is done at school that means I only end up going in for his reviews or talk to them on the phone and don't have to be so involved in that and that is one of the great things about being at the school, he by and large gets what he needs, rather than me being at every appointment. (Wendy, mother of Luke, 11, CHARGE).

Wendy also expressed her frustration that often she has to take her son to the appointment but in fact the consultant does not really need to see him; Helena and her husband have tried to get around this by organising telephone consultations whenever possible.

8.1.5 Summary

In this section about appointments we have discussed issues around getting an appointment, and the different routes to this depending on the kind of clinic participants were attending. Specialist clinics or clinicians with a specialist interest were often considered very valuable, although distances to clinics and the length of time between referrals could be a problem. The process of making an appointment was not always clear to the participants; and they did not feel their individual needs (sensory impairment, communication requirements, several members in one family attending a clinic, or distances they had to travel) were always taken into account. The complexities of their conditions and therefore appointments, as well as juggling the various factors such as childcare, time off work and school, and travel logistics, meant that participants sometimes had to balance how many appointments they could go to. Although creating long days, most participants who were offered multiple appointments in one day (at specialist clinics), and where additional appointments could be dealt with at school, over the telephone, or by a visit to the home, they preferred this.

8.2 At the hospital

Once an appointment had been arranged the next stage in the patient journey was for the participants to attend the hospital. This section will look at the things that contributed - barriers and enablers - to the patient experience 'at the hospital'. This will include examining specifically travel and transport, an overview of the hospital environment, and then cafés and toilets, before providing a summary of the main themes in this section.

8.2.1 Travel and transport

Travel and transport to and from hospital is difficult for all patients, however for those patients who have sensory impairments it is more difficult, for example they may not be able to travel alone, may find it more stressful negotiating public transport, or may have to use more expensive taxis rather than buses. Sensory impairment is additionally stressful and tiring, and all the things that the rest of us find difficult about hospitals is exacerbated, for example finding one's way around the hospital, dealing with people, waiting, understanding what is being said. Additionally in the case of the specialised holistic services, and the condition specific clinics they were often far from people's homes and necessitated a long drive or journey to get there. Furthermore the syndromes were complex (in most cases) and required multiple visits to clinics over the year.

Difficulties with travel and transport to and from hospitals, was one of the most common areas identified for improvement mentioned by participants. For example Jen (mother of Megan, 18, CHARGE) said, "Inside the [hospital] staff are all approachable and things like that. Just the parking situation!" Similarly Maureen (47, Stickler) mentioned, "Overall I would give [hospital] an 8 or 9 out of 10. The parking's not that great, but other than that..."

For a small number of participants travel and getting to the hospital was straightforward as Kim explained:

...it is so much more accessible because it used to be 1 ¾ hours from home across London with buggy, and Lily, and everything

else, and now it is just a 22 minute journey really. (Kim, mother of Lily, 7, Infantile Refsum).

Likewise Linda (58, Usher) said, “It is about 20-25 minutes in the car. My husband goes with me.”

However, even if travelling to the hospital was relatively straightforward, the car park caused further difficulties. For instance for Sandra (mother of Max, 11, CHARGE) it was an hour in the car to the nearest major hospital, and 20 minutes to the local hospital, a distance that she felt comfortable with, however once at the hospital she experienced difficulties finding a place to park and with the associated costs:

The car parking at (major hospital) is awful. I’ve got a Blue Badge so in a sense it is easier than for most people so I don’t have to pay. I would pay £100s if I didn’t have a Blue Badge! The amount of times we go! It’s awful. There are very few spaces, for anyone, let alone someone with a Blue Badge. (Sandra, mother of Max, 11, CHARGE).

Jonathan’s (12, CHARGE) mother was flabbergasted by the £14 car park charge incurred at one of her son’s appointments. Likewise Jen commented:

The hospital car park is actually dearer. I park over the subway – there is another one there – it’s 70p an hour. At the hospital it’s £2, £2.50 an hour. For some families, if they don’t know the area – it’s hard and it’s costly, and if they can’t claim it back. The hospital car park I think it’s stupid what they charge... (Jen, mother of Megan, 18, CHARGE).

Chloe (11, Stickler), when asked about improvements to the condition specific clinic that she attended suggested, “I think the car park should be cheaper.” Her mother Harriet (38, Stickler) also agreed calling the situation “ridiculous”. Harriet went on to suggest that public transport might be an option in some circumstances but for her family - a mother and two children all having examinations and dilating eye drops, as well as another daughter, a car was needed to transport everyone, but even so the situation was described as “quite stressful” for her husband.

Nevertheless Jack (27, Wolfram) was very enthusiastic about the car park at one of the hospitals that he attends. However Jack's experience at his local hospital demonstrates that participants were often drawing on experiences from many different hospitals when reporting their experiences, but also there may be very real consequences for patients if there are difficulties in relation to travel:

The car park is amazing cos you don't need to pay for the blue badge, you press a button on a camera and they open the gate when you show your blue badge. Compared to [local hospital], [local hospital] is pathetic – queuing and queuing and by the time I was getting out it was too late for the toilet and I wet myself. Yea [at hospital] they have a whole floor full of disabled bays so you are never stuck for space. (Jack, 27, Wolfram).

For other patients the long journey times and early starts were the problem as Kevin (guardian of Isla, 8, BBS), Jeff (42, Usher), and Alice (17, Stickler) explained:

Cos it's a 4 hr journey there, 4 hour journey back. That's 8 hours. And an all day clinic. Last time we were there was 9-6, so that's not going to be much good to me. (Kevin, guardian of Isla, 8, BBS).

I have an early appointment so I have to leave about half 4, 5 o'clock in the morning. So then when I get to London if I need to see the doctor, or have a low vision assessment, I'm there about 5 hours. So it's a long day. (Jeff, 42, Usher).

It's a bit exhausting really. Spending 4 hours in a car and then spending 4 hours in a waiting room, and then another 4 hours in the car. It does seem a bit pointless when there's nothing wrong. I haven't had any problems in my eyes in the last few years. (Alice, 17, Stickler).

Alice's comment as to whether it was worth all the travel and waiting around for a check-up when all appears well was repeated by Ellen (38, Stickler) speaking of the care for her daughter Maisie (6, Stickler), "Overall [hospital] is alright, it is a drag and it feels like a massive amount of time but then at least then everything is fine." It is the risk of not going and being told, "actually your daughter's eye detached" that

compels Ellen to attend the clinic with her daughter. This weighing of different factors, such as clinician expertise, hospital location, and the inter-personal skills of the hospital staff, come into play when patients and their families decide whether or not to attend a particular clinic or hospital.

It was not just the distance or the fact that it takes a long time to get to the hospital, for some of the participants the act of travelling caused discomfort. For example travelling for an extended period of time gave Paul (49, Alström) pains in his coccyx. Similarly Vera (83, Usher) found travelling hard and had to spend a night in a hotel before her appointment in order that she was in a good shape to attend her appointment:

With my age as well it makes travelling really difficult. Perhaps if I was younger it wouldn't be so bad. But I need a rest before and after I've been [to my appointment]. (Vera, 83, Usher).

For Wendy's son Luke (11, CHARGE) it was not the fact that the Tube station was far away that caused them both difficulties but because of Luke's mobility the distance away for *him* was far. Wendy was speaking about her experiences at one particular hospital where parking is very limited and all patients are advised to come by public transport. Becky (mother of Josh, 13, CHARGE) also commented on this suggesting that this just added an extra layer of stress and anxiety on to the day for her son:

Getting to the hospital from the tube is fine. We can walk that distance no problem. Parking at [hospital] is a nightmare. We just don't do it really unless he's in for surgery and we get a special car parking pass but it is horrendous to try and park up there. I have to say that most of the time it is ok, it is just when the tube is busy it makes Josh very anxious. And not for a pleasant journey. When there are delays or tube cancellations it is awful. (Becky, mother of Josh, 13, CHARGE).

This added level of anxiety and stress arising from travel complications was also mentioned by other mothers as Helena and Jen demonstrate:

It can be a long day and you've always got the car parking considerations. There's never enough car parking. We've got a blue badge so we can park in the residents' parking bays but that's if you can find one, and then if you end up parking on a double or a single yellow you've only got 3 hours. They might tell you that your appointment is at 10 o'clock but you're still there at half one waiting to see somebody. It's really stressful. The whole thing in general tends to be quite stressful. (Helena, mother of Rosie, 4, CHARGE)

Sometimes you can wait a good hour to get seen. And that's frustrating if you have gone in the car cos you've parked it in the car park... Which way to go – do you put the extra hour on? Sometime you might put three hours in and then you're seen within half an hour and you think what was the point of that! They could do with a 'pay after' car park. So at least then you know what you've been. (Jen, mother of Megan, 18, CHARGE).

Three participants Jeff (42, Usher), Doug (60, BBS) and Basil (57, Usher) spoke of their experiences of using patient transport for getting to hospital. Whilst they recognised the usefulness and were grateful for it, it involved a lot of waiting around:

It's in a people carrier. It picks up 2 or 3 other patients en route or there's 2 on the bus already. It fits three or four people in. You have to be ready hours before and hours after but it's fine and you get there and back. It's the only way I can do it. (Basil, 57, Usher).

One thing that you have to wait long for is the ambulances. To get there and back and sometimes they don't turn up... ..You have to be ready two hours before your appointments. And then they can come any time within those 2 hours. So you've just got to hang around and wait. It's a bit frustrating. (Doug, 60, BBS).

Reliance on Patient Transport and the strict regime meant that Jeff, whose low vision assessment took 2 hours instead of the anticipated 30 minutes, and he had to leave before seeing the consultant as he relied on Patient Transport to make the journey back home. Likewise, it was observed as part of the accompanied clinic visit that Basil arrived 45 minutes late for his appointment as the Patient Transport had arrived late. This left Basil feeling very agitated and distressed, particularly as

he had something to do in the evening and was unsure whether he would have time to see the consultant before he had to leave for his next engagement – luckily for Basil it appeared that he was moved through the queue quite quickly and was able to be seen by both the registrar and the consultant.

Angie (43, Stickler) said she was unable to access Patient Transport. Her GP and local services said that it was the role of the specialist clinic, and the specialist clinic suggested it was the job of the local NHS Trust to provide the transport, which ultimately left Angie in the situation that she may have to cancel her next appointment at the specialist condition specific clinic as she has no way of getting there.

Some participants were able to claim for transport costs, which was very useful, as Ellen (38, Stickler) explained, “We are lucky in that we have tax credits so we claim mileage and parking. But it would be expensive [otherwise].” For Phillip (28, Wolfram) if he had not been able to claim travel costs it is unlikely that he would have been able to attend the specialist holistic service. However the procedures involved in being reimbursed were quite complicated and inflexible as Alexa explained:

It’s very bureaucratic. If you don’t have the right pieces of paper, then it’s just a straight ‘no’. There’s no kind of listening to your individual circumstances, you know if your documentation doesn’t fit the criteria, they don’t care if you have spent £50 getting there. (Alexa, 27, Stickler).

The procedures varied from hospital to hospital and were also dependent on which service was being accessed and where the funding was coming from. For example when Paul (49, Alström) attends the specialised holistic services, “We get reimbursed for the taxis and trains. That’s from the CCG, something to do with the NHS.” However when he attends a local hospital, for other related services such as hormone injections, feet checks, and blood pressure monitoring, on average 12 times a year he has to pay £7, each way, for a taxi, every time he attends. As Paul commented, “I don’t like it.”

8.2.2 The hospital environment

The hospital environment, incorporating lighting, signage, noise levels, and ease of access is very important for promoting mobility and communication – two of the three key areas that have been identified as having specific significance for people with a dual sensory impairment.

As with many aspects of the hospital and clinic experience, the participants had varying opinions on the environment and this was dependent upon their own particular needs, as well as the hospitals that they attended. For example Leo (17, CHARGE) when asked about the hospital lighting said, “It’s fine.” When Basil (57, Usher) was asked the same question he responded, “I don’t know because I am registered blind. I suppose you could say not applicable.” Meanwhile Jen (mother of Megan, 18, CHARGE) replied, “It’s ok but sometimes there is the glare with the bright lights. That can affect her eyes. But she’s ok.”

Glare was one issue that was reported in relation to lighting. Another was the brightness as Jean and Jeff explained:

It’s a bit dim. It’s alright in the actual waiting room for the ophthalmology department but walking through the hospital it is not very good for me. (Jean, 67, Usher).

It’s dark...It’s not very good. It depends on the weather. Whether it’s light or sunny outside. Walking into the building takes a bit of time to focus. (Jeff, 42, Usher).

However for Vera (83, Usher) the lighting at the hospital she attended was appropriate for her, “you’ve got the very bright walls and the strip lighting, that’s good.”

Contrast, or lack of it, was mentioned by Nina in relation to her appointments at an eye hospital:

To be honest the whole thing has got a sort of grey colouring.... it’s kind of a bit too monotone. The walls and the floor don’t contrast really which is kind of surprising really considering the type of hospital that it is. (Nina, 46, Usher).

As identified previously, when there was an example of less accessible environments in relation to vision in an eye hospital or ophthalmology department, participants felt more negatively about the situation than in another type of hospital as Rachel also remarked:

I would change the lights definitely. I don't really understand it because lots of people going into the eye hospital will have eye problems and that... (Rachel, 40, NF 2).

Opinions on the hospital environment differ from patient to patient as the two examples from Alice and Dave who both have Stickler and were talking about the same condition specific clinic:

It's too bright. It's all very white and clinical. It's a bit too bright. (Alice, 17, Stickler).

...the main clinic now is the modern annex on the side of the building which has been designed with good lighting and signage and I think that's quite good. (Dave, 43, Stickler).

This being so, what was useful for participants was the opportunity and willingness from staff to adapt the environment and make it more accessible as necessary. In the case of Jessica (21, Wolfram) and Phillip (28, Wolfram) it was the fact that there were staff and volunteers available to help show them the way when necessary. Kevin (guardian of Isla, 8, BBS) identified that anti-glare film had been placed on the windows of their local hospital which "takes the glare off everything." For Simeon (46, Usher) if hospitals were able to use 'natural' daylight bulbs rather than the traditional sort of bulbs, that would be very helpful to him. It was observed in Moorfields Eye Hospital that the distance in metres to certain departments was written on the walls. For Rachel the opportunity and space to rest was invaluable:

Before [the lighting] was fine, it was nice and bright for a deaf person. But now with my eyes, it's too bright. It makes me feel sick because they are bright. I am very very sensitive to light. It makes me feel sick and my head goes wuzzy. Sometimes I go into a room and turn the lights off and I just sit and wait in the dark. (Rachel, 40, NF 2).

Rachel's comments serve as a reminder that not only might patients' needs change over time but the unique nature of dual sensory impairment means that sometimes the simplest and most obvious solutions are the most appropriate.

Overall in the hospital environment patients considered the noise levels acceptable, however as we shall see in the next sections it is mainly in the waiting room or consultation room that noise became an important issue. As Jean (67, Usher) commented, "[noise] doesn't matter so much walking through the corridors but when you get to the ophthalmology department it is too noisy."

Clear signage, in relation to font, size, contrast, and positioning, is an important feature in creating an accessible hospital environment, as are clutter-free, smooth (involving no unnecessary changes in flooring) walkways and corridors with no trip hazards. The environmental audits were a useful tool in helping to demonstrate both good practice and room for improvement in relation to signage and mobility within the hospital:

Table 2: Environmental Audit – Hospital, general signage

HOSPITAL	General signage
1)	<p>Very large, good contrast, overhead signage walking through main doors - blue with white writing.</p> <p>Main sign indicating all wards and departments when walking into hospital very poor – blue text on clear plastic signs overlaid artwork. Very little contrast and difficult to see as positioned over the artwork.</p> <p>No braille or embossed signs.</p>
2)	<p>Large signs, numerous, clear font.</p> <p>Blue with white writing. Good contrast.</p> <p>No braille or embossed signs.</p>
3)	<p>Large, good contrast signage at entrance.</p> <p>Too much signage, not easy to pick out the information need.</p> <p>Signage to clinic was not well contrasted.</p> <p>No braille or embossed signs.</p>
4)	<p>Signs to inside the hospital were generally black with fluorescent yellow writing and trim.</p> <p>Signs from outside the hospital blue with white writing - large, good contrast but if you had been looking out for 'blue and white' signs, or vice versa, could be confusing.</p> <p>No braille or embossed signs.</p>
5)	<p>Clear, good size, good contrast purple signs with white lettering.</p> <p>No braille or embossed signs.</p>

Good examples of clear and visible text were seen in all of the hospitals. Clearly readable text included blue backgrounds with white writing, black background with fluorescent yellow writing and purple background with white writing. However, in hospital 1) some signage was very difficult to read, for example blue text was on clear backgrounds which overlaid art work, and in hospital 3) the multitude of signs, especially near the lifts and at the entrance meant that it was difficult to locate clinics or departments and the corresponding arrows. Furthermore in clinic 4) although signage had good contrast and was of a good size there were two different colour schemes (blue and white outside the hospital – the more direct route to the clinic, and fluorescent yellow on black within the hospital) which could be confusing if you were looking out for ‘blue and white’ signs or vice versa. None of the hospitals had braille or embossed signs.

Table 3: Environmental Audit – Hospital, movement within corridors

HOSPITAL	Movement within corridors
1)	<p>Cluttered corridors – beds, equipment, bins. Not good for someone who trails, or needs a wall nearby.</p> <p>Artwork – professional and children’s on the walls, makes friendly and welcoming. No tactile artwork.</p> <p>One sudden change in flooring colour which gave the impression of a step but was not a step- caused the researcher to stagger.</p>
2)	<p>To outpatients clinic, wide, bright and airy.</p> <p>Clutter free corridors, not far to walk.</p> <p>Lots of seating along the way for people to pause.</p> <p>Not able to trail along walls as very wide and seating in the way.</p>
3)	<p>Confusing building with numerous staircases and ways to get to different places.</p>

	Clutter free corridors but busy.
4)	Clutter free corridors. Grab rails. Seating at various points. Different artwork to make attractive. Some areas were dark and dingy.
5)	Clutter free corridors. Good contrast use of contrast on the floors and walls. Making use of natural light where possible. Art on walls – paintings/transfers. Colourful but not overwhelming.

Hospitals 2), 4), and 5) had clear clutter free corridors which were easily navigable. Although the corridors in hospital 3) were clutter free they were very busy with lots of people walking around which people with sensory impairments may find more difficult to navigate. In hospital 1) medical equipment, laundry bins, and beds were in the corridors. Artwork on walls in hospitals 1), 4) and 5) made corridors seem more welcoming. However although some areas in hospital 4) had beautiful and engaging artwork other areas were quite dark and dingy. Seats on the route in hospitals 2) and 4), as well as grab rails in hospital 4) gave the opportunity to pause. Corridors that were too wide or too cluttered also presented problems for trailing, as seen at hospitals 1) and 2).

All routes were step free if people chose to use the lifts, however in hospital 1) there was one area where the colour of the flooring changed suddenly and caused a possible trip hazard, although there was in fact no change of level.

Although there might be clear accessible signs the information contained within them needs to be appropriate as Louise explained:

[Hospital] is not remotely accessible. They do things like, they send you a letter saying “report to the eye unit on arrival” and you get

there and it is called Ophthalmology. Well I know that ophthalmology is the eye unit but other people might not. There is no sign that actually says eye unit. (Louise, 41, Usher).

Some participants were moving around the hospital from memory, so signage was not so important, however there was a fear that if clinics moved they might not be able to find their destination:

I now work with photographic memory to get to the clinic and things like that because when I used to go, I used to go with my husband so I know what I were doing. I think if they were to change it, the location then I would struggle. (Donna, 46, Usher).

When moves had taken place there were difficulties and frustrations:

The car park is really confusing, I couldn't work out where to park and the hospital is huge. We get lost and every time we go there they have moved the clinic. We used to park and know where we were going and they moved the clinic and we couldn't find it and my Dad now is blind and can't see. It was the biggest nightmare in the world! It is like the hugest hospital I have ever been in in my life! (Roxy, 29, Stickler).

One time they moved offices and before he used to look out the window at the 20 traffic lights and he was in heaven, and then they moved in a stairwell, there were no windows for a start, and because I had gone unprepared for the move, and he was so cross at the change, I think he cost them over £300 worth of damage easily! (Sandra, mother of Max, 11, CHARGE).

In these instances there might not be much that the hospital can do, in that the changes have to go ahead, however what is crucial is that the hospital provides patients with appropriate (and accessible) information about these changes. An example of this relates to the young people who were transitioning or had transitioned from specialised holistic services at Birmingham Children's Hospital to the adult specialised holistic services at the QE Hospital in Birmingham. 'Transition Days' were arranged when they had been to visit the QE hospital and met the staff and got to know the hospital environment.

8.2.3 Cafés

Helena (mother of Rosie, 4, CHARGE) describes “the three really important things when you’ve got small children” “Food(s?), toilets and playing space!” All three of these elements will be looked at in this report, beginning first in this section on cafés, before moving on to ‘toilets’ in the next, with ‘spaces to play’ to be discussed in section 8.3 ‘At the clinic’

Participants were asked for their opinions on the cafés and the refreshment facilities that were available at the hospitals that they attended. Not all the participants used them but for those who did a large number of participants reported that the refreshment facilities were ‘fine’, as Gordon (34, Usher) commented, “There is café there. I have been there once and it was fine.” Similarly Rachel (40, NF 2) noted, “[the café] is ok.”

For some participants, particularly those who were caring for children with complex needs they would not have refreshments themselves but would try and make sure that their children ate and drank at regular meal times, often bringing their own food to the hospital in order to manage this, as Kim explained:

I would probably take Lily’s lunch with me and just sit with her quietly and I wouldn’t bother to have anything to eat or drink myself. I would just wait for her, and after the appointment is finished I would go back to get her back into the school routine. To have lunch at school. (Kim, mother of Lily, 7, Infantile Refsum).

For Harriet (38, Stickler) and her family it was necessary to bring food from home as the prices in the cafés were too high, an attitude shared by her son Ollie (6, Stickler) and daughter Chloe (11, Stickler).

Researcher: What would you change about hospitals?

Ollie: Food.

Harriet: Yes ‘cos it cost a fortune when you are in hospital.

Chloe: It was two quid for a sausage roll and it’s about 80p at my school dinners.

Whereas for other participants the opportunity to get something in a café added a high point to an otherwise not so pleasant day, as Becky, Louise, and Jonathan explain:

You can take your own food in there if you want, but we quite often stop and have something to eat. If he has the day off school, we kind of make a bit of a day of it. (Becky, mother of Josh, 13, CHARGE).

Again I am really lucky that (eye hospital) has a Costa in it. So the coffee is very good. This year I have had an awful lot of appointments. There has been a deterioration in my Ushers this year. And my husband has taken 10 days of annual leave to take me to appointments. We are not having a holiday this year because when we go to a hospital appointment, we have a day out. If we go somewhere that has good coffee that's a really good day out. That's how we see it. Even if it is a hospital they have got Costa. You know. It is a bit sad isn't it! (Louise, 41, Usher).

Researcher: What do you especially love about hospitals?

Jonathan: The check-ups and part of it is the coronation chicken sandwiches that I like to get afterwards... (Jonathan, 12, CHARGE).

As well as expense being one of the difficulties in relation to refreshments, the café was also sometimes quite a distance from the clinics the participants were visiting. For example at one of the hospitals the café was around 700 paces from the main clinic area, and at another of the hospitals it was 4 floors from the clinic to the main café, requiring some travel. . For Holly (49, Usher) at one of the hospitals that she attended the café was down two flights of stairs. In order to ease some of the distances involved in getting to the cafés sometimes smaller cafés or kiosks were located closer to the clinics, for example in the main outpatients clinic at hospital 1) there was a small kiosk selling drinks and pre-prepared foods whereas downstairs in the basement was a larger café selling a wider range of food and drinks, including hot food. At the ophthalmology outpatient clinic squash and water was available in the clinic area. Wendy (mother of Luke, 11, CHARGE) found this particularly

useful as her son is not a “big drinker” and it is “important to get fluids into him throughout the day”, particularly in “hot, stuffy, waiting rooms like this.” Similarly at one of the hospitals that Donna (46, Usher) attended, drinks were provided in the waiting room by someone with a tea trolley.

During the environmental audit in clinic (i) it was observed that a free lunch, for all patients and those accompanying them (parents, siblings, grandparents, interpreters, and intervenors) was provided. The lunch not only provided sustenance, but gave families the opportunity to talk to each other and provided a break in an otherwise quite hectic day. The clinic co-ordinator from the charity associated with the syndrome was also present and was able to answer any questions. (Further information on the clinic co-ordinators can be found in section 8.3). Some clinicians also ate at the lunch, allowing the opportunity for some informal interaction between medical staff and patients. Conversations with clinic co-ordinators of the specialised holistic services at Birmingham Children’s Hospital and the QE Hospital in Birmingham indicated that such lunches also take place at these clinics.

Some of the problems related to refreshments stemmed from mobility and lighting issues, demonstrating that the environmental aspects of hospitals cut across many facets of the patient experience. These will be discussed in further detail in the section below, but in relation to cafés (at two different hospitals) Jean (67, Usher) and Jeff (42, Usher) had specific difficulties due to the environment and had to rely on another person (in this case their spouse) to access the café:

As we entered through one of the doors there was a small café there. I just sat down and let my husband do it. I don’t think I would even attempt it because the lighting levels are inconsistent – they might be light in one place and dark in another. Then you’ve got the lights coming up from the refrigerator displays which are quite bright. So all the changes in lighting levels are quite confusing. There are people queuing and then there are barriers. So you don’t know if you are going to walk into a barrier. You are not sure where you are going to go to pay. So I don’t even attempt it! (Jean, 67, Usher).

Jeff: (Wife) takes me into the café but it is very, very dark in there. I go and sit down and (wife) goes up to the counter and gets herself a cappuccino or coffee.

Wife: There's too many chairs and that in there for Jeff to go up and have a look. It's got quite a few bollards.

Jeff: Very small.

Wife: They've got quite a lot of tables and chairs in there. It's not really... Considering it's an eye hospital I'm quite surprised that there was that many tables and chairs in there. I understand it's small, they got a lot of people and everything, but I just found for an eye hospital the café's really was a bit too overcrowded with all the tables.

Jeff: And it's too dark. Brown walls.

Wife: Yeah. It really was too dark in there.

Similarly Vera (83, Usher) who used a wheelchair due to old age, rather than any specific aspect of having Usher, found the café too crowded and her daughter was unable to push her in wheelchair as there were too many tables and chairs. Mobility issues cut across impairments and disabilities, and are not just specifically related to the condition under investigation.

Jeff's wife remarks "I just found for an eye hospital the café really was a bit too overcrowded with all the tables" returns us to the point that participants were particularly frustrated or disappointed if a specialist eye hospital did not meet the needs of its visually impaired (or dual sensory impaired) patients.

The table below provides an overview of the main café at the five hospitals where environmental audits were undertaken:

Table 4: Environmental audit – Hospital, (main) cafeteria

CLINIC/HOSPITAL	Cafeteria (main)
1)	<p>Dark – natural light at only one end</p> <p>Chairs easily moved around</p> <p>Limited options for food</p> <p>Menu in very small print</p> <p>Staff not helpful</p>
2)	<p>Good variety of food</p> <p>Helpful staff</p> <p>Lots of seating, easily moved around.</p> <p>Good lighting, main use of natural light.</p>
3)	<p>Main café was closed at 2.30pm on day of visit</p> <p>Smaller café was very dark and tables crammed together. Would be hard for a wheelchair user to pass through.</p> <p>Friendly staff.</p> <p>Menu on board behind the till – poor contrast and far away</p>
4)	<p>Dark and quite warm (cooler, lighter area was staff only)</p> <p>Very busy and noisy</p> <p>Lots of food choices</p> <p>Helpful staff</p> <p>Menu in small print</p>
5)	<p>BBC news on TV with subtitles and sign language</p> <p>Modern but the sweeping lines and low ceilings</p>

	<p>could be a bit confusing/annoying for some people</p> <p>Slightly dark – lack of natural light</p> <p>Small print signs telling you what the food was – wipe boards where the food choices could have been written in a good size and with contrast were not in use.</p> <p>Chairs easily able to move around.</p> <p>Tables, chairs, floor, good contrast.</p> <p>Helpful staff.</p> <p>Cutlery and trays good contrast.</p> <p>Large play area to one side – not intrusive to the restaurant</p> <p>Good variety of food</p>
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The environmental audits provide us with examples of both good practice and places where things could be improved, as well as a snapshot of the experiences for patients attending these hospitals. Once again it is sometimes the small things that a hospital can do to provide better access for its patients, for example at hospital (5) the TV showing the news had subtitles and sign language and there was good contrast between the trays, cutlery and tables. None of the menus were available in large print, and at hospital (1) the menu was in very small print. At hospital (5) there was a wipe board where the menu could be displayed in large print however this appeared not to be in use. The chairs and tables in hospitals (1), (2) and (3) were movable which can be useful for people who have mobility difficulties, or who need to sit in a particular configuration to support communication. Cafés were also busy, particularly at hospital (4) which made moving around difficult. Lighting in the cafeterias was general dark, however in hospital (4) it was noted that there was a cooler, brighter, and less crowded area however this was only available to members of staff. The majority of staff were however friendly and were happy to assist customers.

8.2.4 Toilets

For a lot of participants the toilets were 'ok' and readily accessible, as Mohammed (9, BBS), Rachel (40, NF 2), and Phillip (28, Wolfram) commented:

Yeah, they're ok. They're not really far from us. (Mohammed, 9, BBS).

Yes they're all right. (Major hospital) is new. It's a new building, so it's all really good. Lots of space and toilets. (Rachel, 40, NF 2).

Yup, they seem to be ok – there usually seems to be plenty of them. I usually use disabled toilets, because they are wider and if I have to change my catheter I've got the space to do that. (Phillip, 28, Wolfram).

Alternatively a smaller number of participants were happy to ask for assistance, for example Doug (60, BBS) said, "Yeah I just ask one or another. Someone'll show me the way." Similarly Basil (57, Usher) commented, "If I need the loo I make sure that I am near the reception desk and I know which way to go. Or I ask someone to take me to the toilet and back."

However a couple of participants mentioned that they did not use the toilet at the hospital "I've never been" said Gordon (34, Usher) and similarly Donna (46, Usher) laughed, "I have always worked on the mental attitude that I don't go to the toilet in the hospital." It is interesting to wonder whether participants just happened not to need the toilet, or whether it was a conscious decision not to go, and this was a strategy that worked for them.

Some participants found it difficult to locate the bathroom as signage was difficult to see, as Simeon (46, Usher) commented, "these little [signs] I don't see them." Simeon suggests that all toilets should have "big man, big woman, big baby" signs on them. Jean (67, Usher) also agreed that the toilets were poorly signposted, however she was unsure of what improvements could be made:

I don't think they are particularly well signed but I don't know how you would make them better signed for someone with tunnel vision. (Jean, 67, Usher).

Although there might not be anything specific that could be done to support Jean's needs the recognition that it may be difficult for her and that she has to rely on her husband to point out the toilets to her, is an important contribution to her experience of hospitals and clinics.

For Nina (46), Jeff (42), and Vera (83), who all have Usher, the issue of the lack of colour contrast in the bathroom was troublesome, as Vera, explained:

The toilets were difficult because it was all white. The tiles were white, the basin was white. The walls. You are really having to feel all your way around to figure where everything is. Not ideal in a toilet. And you don't know what you are putting your hand in! And then I have trouble finding the door handle and how to get out. I just had to bang and my daughter came in and got me. Not nice. (Vera, 83, Usher).

For Nina the difficulties with the lack of contrast in the toilet, describing "trying to find the thing to flush the bloody loo is my bugbear!" was even more frustrating as, "in a hospital you'd have thought that they would have got that thing sorted." This belief that things should be better in a hospital than in everyday life and the ensuing frustration when this was not so, was a common theme of participants' experiences.

Nina had a suggestion that she wished to share as to how hospitals could improve their access for patients:

They could even produce a patient leaflet saying to you; all the toilets here have red doors, and they are all of the push mechanism, and even have a picture of the loo, just so when the person goes there they know what to expect and it's just one less thing to be anxious about. (Nina, 46, Usher).

Nina's suggestion shows that it might be useful for hospitals to speak to patients and ask what sort of information they might require and that although toilets might seem an insignificant part of the hospital

experience for many patients, for a number of patients it may be a source of anxiety and apprehension.

Nina's comment of "it's just one less thing to be anxious about" links to Chloe's comment about needing the loo at the hospital but the accompanied concern that she might miss being seen:

The [toilets] are sort of [easy to find] but they are a little bit far away from the waiting room, so if you need the loo and you have to walk and you don't want to as you might miss the Doctor. (Chloe, 11, Stickler).

As will be discussed throughout this report, low levels of anxiety, often centred around certain key points, were a typical feature of a participant's experience of hospitals and clinics.

It is interesting to note in the example about colour contrast in the toilets that all the participants mentioning this had Usher and were speaking about their experiences of a particular hospital. This may suggest that it was an 'Usher issue' and specific to one hospital, nevertheless it highlights the difficulties that some of the participants in the research project faced, and the specific needs of this particular patient cohort. Similarly Sandra (mother of Max, 11, CHARGE) identified the specific needs of her son Max in accessing the toilet:

If he gets stressed he'll tend to poo. Which can be a bit difficult because you think, should I go because then I'll miss the appointment, and I've been waiting for 2 hours. And also there's never anywhere to change an older child that requires lying down. There's nowhere. In most places you could probably ask. There's often not a room free. At (local hospital) there is. They'll provide a room. But it's disgusting if not. You're on the floor. It's not ideal. At the eye hospital as well they don't have anywhere to get changed and the toilet is miles away from the waiting area so it's not as if you think you'll be in and out in minute, you have to go down a few corridors. (Sandra, mother of Max, 11, CHARGE).

Sandra was aware that although they were specific needs, her son was not the only child with such needs and she was frustrated as to why,

even in a hospital, these needs could not be met. (Sandra felt pleased that at least at her local hospital a room was set aside for changing children who needed to lie down – however this was not a designated bathroom). Out of the five hospital environmental audits it was noted that in only one of the hospitals (5) was there clear signage directing people to a toilet with a ceiling hoist and changing bed.

The environmental audit also looked at the toilets to explore how the environment in the bathrooms supported the needs of people with sensory impairments and potentially complex medical needs. The environment of the nearest toilet to the clinic, as well as any disabled access toilets was recorded.

Table 5: Environmental audit – Clinic, toilets

Clinic	Toilets
i), ii), iii)	<p>Individual toilet with door on to the corridor</p> <p>Good contrast between toilet seat and floor, and sink and walls.</p> <p>Emergency pull cord.</p> <p>Space for an accompanying person.</p> <p>Toilet door opens on to corridor so not suitable for someone who needs the door kept ajar.</p> <p>Light, bright and airy.</p> <p>Not enough space for a wheelchair user to transfer.</p> <p>No toilet with hoist or grab rails nearby.</p>
iv)	<p>3 cubicles in toilet block, with sinks outside the cubicles.</p> <p>Cramped, hot and dark</p> <p>Not much space for an accompanying person.</p> <p>Cubicle door opens on to sink area so suitable for someone who needs the door kept ajar.</p> <p>Not enough space for a wheelchair user to transfer.</p> <p>No toilet with hoist or grab rails nearby.</p>
v)	<p>Individual toilet with door on to the corridor.</p> <p>Bright and light.</p> <p>Emergency cord.</p> <p>All white sink and toilet, white floors, green walls.</p>

	<p>No space for an accompanying person.</p> <p>Toilet door opens on to corridor so not suitable for someone who needs the door kept ajar.</p> <p>Not enough space for a wheelchair user to transfer.</p> <p>No toilet with hoist or grab rails nearby.</p>
vi)	<p>4 toilets (including one disabled access toilet) in a large block - had grab rails and space to transfer but no signage inside the bathroom as to which toilet it was.</p> <p>Blue doors, grey floors, white toilets and sinks – lack of contrast.</p> <p>Good lighting.</p> <p>Space for accompanying person and suitable for someone who needs the door kept ajar.</p> <p>No toilet with hoist nearby.</p>
vii)	<p>Single, unisex and disabled access toilet, with door opening on to the corridor.</p> <p>Grab rails, just enough space to transfer.</p> <p>Emergency cord</p> <p>Toilet and sink well contrasted.</p> <p>No toilet with hoist nearby.</p>
viii)	<p>3 or 4 toilets in a block.</p> <p>Bright and airy.</p> <p>Flooring white/grey with white sink and toilet</p>

	Enough space for an accompanying person
	No toilet with hoist or grab rails nearby.

Once again we saw that the hospitals met some of the requirements of people with sensory impairments and complex medical needs however not all of them. Lighting was good in all of the toilets except at clinic (iv). As mentioned by patients above, good contrast between the sink, walls, and toilet seat is important - this was only achieved at clinics (i), (ii), and (iii) (which was in fact the same space but used by different clinics) and at (vii). For those patients who might need an accessible toilet they were often further away from the clinic than other toilets which exacerbates the anxiety of missing your name being called. There were no toilets nearby that provided a hoist or changing bed for patients who require this. In clinic (vii), where there was an accessible toilet close to the clinic, it being inside the main toilet block meant that it was harder to negotiate for people with a visual impairment. Some of the toilets were very cramped and did not allow another person to accompany the patient, if this was needed. Also as seen in clinics (i), (ii), (iii), and (vii) because the door opened straight on to the corridor it was not suitable for someone who needed to 'keep an eye on' someone and keep the door ajar whilst they used the toilet independently.

8.2.5 Summary

In this section we have examined travel, the environment, and hospital facilities.

Difficulties with travel to hospital are a problem common to most people attending hospitals but are exacerbated by sensory impairment, and having blue badge parking did not always alleviate this. Because of the rarity of their conditions, and the specialisms of hospitals, travel often took a long time and was expensive, and the journey itself caused stress and difficulty. In looking across different hospitals, some had good lighting and minimised glare, some had good colour schemes which identified areas and furniture, some had clear, good sized signs, some

avoided clutter in areas where people were moving, but none managed all of these. Once within the hospital, cafés were a key part of the facilities for some people, but they could be expensive and sometimes they were a long way from the clinic, they were not well lit, and they did not provide for sensory needs, for example with braille or large print menus. Toilet facilities are obviously very important when people are waiting for some time or have multiple appointments. Some found that they were easy to get to, but others found that sensory issues in both signage, and in bathroom furniture, made it difficult for them to be independent.

8.3 At the clinic

Having considered the first and second stages in the patient journey – the appointment and at the hospital – this section will examine the participant experience at the clinic incorporating the reception and waiting areas and the waiting room, before drawing on the role of specialist co-ordinators at the clinic.

8.3.1 Reception

Having made their way to the clinic area, the next thing that the participants had to do was to check in at reception. In some clinics there were touch screens to allow patients to check in automatically. Jean and Gordon, speaking of two different hospitals, questioned the appropriateness of this in an ophthalmology clinic:

...we noticed last time that you can register your presence as soon as you get in there. They've got a lot of computerised registrations. Which is fine if you can see them, which I can, but if you couldn't I think you'd have to just walk and find it! (Jean, 67, Usher).

...I found it comical that the hospital, when you walk in, when you get to the check in desk, they don't do check ins any more, it's a machine you've got to do yourself, in an Eye Hospital, where there are people who are blind, and I thought that's hilarious! It's taking the piss! Now my eyesight, because I wear contacts, I can see what's right in front of me, but someone who's blind is not going to be able to do that! (Gordon, 34, Usher).

Touch-screens to register your presence were also seen at clinic (v), however there was also a desk with a person behind it so people could communicate face-to-face if they so wished. Face-to-face communication is often important for people with sensory impairments, particularly for those who rely on lip reading, or for those who would not be able to either see or operate a touch-screen.

Despite clear communication being important for the participants in this research project a number of participants identified that their communication needs were not being met by reception staff:

...they don't do face-to-face contact. They start saying something and then look down at the computer. You don't get the details so you have to say what did you say, I didn't quite catch that. So it gets confusing with me on that one. (Jeff, 42, Usher).

I think the staff in the reception, I know they have a job to do but I think a little bit of re-training programme and thinking "not everyone is the same, not everyone is like them. There are people with disabilities and they don't see". (Linda, 58, Usher).

You go to the desk and they don't take their eyes off the computer so they don't make eye contact with you so you can't lip read them. So you are going.... "hello I am a lip reader" oh right.... it is rude never mind being inaccessible. Just look up and give attention to the person who is talking to you. You have no idea what accessibility requirements that person has got. You know and you are just making assumptions that they can hear ok and that they see ok. (Louise, 41, Usher).

In the accompanied visit b) the participant found communication with the reception difficult as "The receptionist did not look up." Donna (46, Usher) and Chris (31, Norrie's Disease) noted that communication was better at audiology clinics, however to some extent this was presumed as "that is more communication related" (Donna, 46, Usher). In accompanied visits (d) and (e) the receptionist kept having to leave the desk so the patients had to wait in a queue to be seen.

Caroline had a unique way to ensure that the staff at the hospital communicated appropriately with her, she went by herself as she found that if she went with her husband or children they would speak to them and not her:

Because I can understand better if I am on my own [they make more of an effort to communicate with Caroline]. I am not part of the Deaf community, I am in a half-way house. If you take someone with you, that is who they speak to. (Caroline, 45, NF 2)

Deferring to the person accompanying the patient was also observed by the researcher in clinic visit e) and by Marg's (48, Stickler) husband in the interview with Marg.

An environmental audit was conducted in the reception area of eight clinics to investigate factors that led to an accessible environment or contributed to an inaccessible one for people with the syndromes under investigation. We took into account the likely needs of the participants, so looking at factors such as receptionists not sitting behind glass screens, access to a hearing aid loop system, low level desks, and appropriate lighting, as well as any other features which promoted a more/less accessible environment, and observed whether these were made available to patients.

Table 6: Environmental audit – Clinic, reception

CLINIC	Reception
(i)	<p>Low desk, wheelchair accessible, no glass, friendly welcoming receptionist took researcher to waiting room</p> <p>Met by clinic co-ordinator.</p> <p>No loop information.</p> <p>Standard lighting – neither very bright nor very dark.</p>
(ii)	<p>Low desk, wheelchair accessible, no glass, friendly welcoming receptionist.</p> <p>Radio playing.</p> <p>Specialist co-ordinator had not arrived</p> <p>No loop information.</p> <p>Poor lighting – lack of natural light.</p>
(iii)	<p>Low desk, wheelchair accessible, no glass, friendly welcoming receptionist.</p> <p>Radio playing.</p> <p>Clinic co-ordinator waiting in reception – greeted patients.</p> <p>No loop information.</p> <p>Poor lighting – lack of natural light.</p>
(iv)	<p>Low desk, wheelchair accessible, no glass, friendly welcoming receptionist. Told to take number and report to a numbered desk.</p> <p>No loop information.</p> <p>Good lighting, cream walls, high ceilings.</p>
(v)	<p>Low desk, wheelchair accessible, no glass, brusque</p>

	<p>receptionist.</p> <p>Small printed sign asking if you have any communication requirements to let reception know.</p> <p>Small loop sign.</p> <p>Bright and airy, good use of natural light.</p>
(vi)	<p>High desk, not necessarily wheelchair accessible.</p> <p>Small loop sign.</p> <p>Desk was often left vacant – receptionist going off to do other things.</p> <p>Bright artificial lighting.</p>
(vii)	<p>Low level desk, no glass, wheelchair accessible.</p> <p>Small hearing loop sign</p> <p>Friendly receptionist but gave wrong info – asked where nearest café was and sent to main one (700 paces) nearby one 250 paces.</p> <p>Bright artificial lighting and some use of natural light.</p>
(viii)	<p>By-passed an interim reception desk (patient's mother knew where she was going and headed onwards)</p> <p>Clinic reception – crowded cluttered desk, in one corner, nurses standing around so that's how we knew to go there – no 'reception' sign.</p> <p>No loop sign.</p> <p>Standard lighting – neither very bright nor very dark.</p>

None of the clinics managed to provide all the things that were identified as making the reception environment accessible for patients with dual sensory impairments. However at none of the clinics were members of staff behind glass, which helped to promote good communication. The

lighting in the clinics varied, and as was mentioned previously not all hospitals can improve upon this, however it is important that those working in hospitals know that this might influence the patient's ability to communicate and move around effectively. In clinics (ii) and (iii) there was a radio playing, and although this might have been useful for some of the patients waiting, it can be an annoyance for those that need a clear background to be able to communicate effectively. Although the environmental audits are one-off snapshots of the hospital environment notes were made with regard to the welcome given by the receptionist as the attitude of a receptionist (and other members of staff) can stay with a patient long after the clinic appointment is over. In clinics (i) and (iii) the clinic co-ordinator was seen greeting patients personally, the positives of this will be examined further in section (8.3.4).

8.3.2 Waiting

Waiting times for appointments was one of the most difficult things for participants, for example Becky (mother of Josh, 13, CHARGE) said, "The worst thing about the clinics is when they are running really late." Linda (58, Usher) commented, "I think the waiting time itself needs to be focused on really. I can be sitting there from the time I check in for well over an hour before I even get called."

However participants were generally understanding when delays occurred as they recognised that it was because an emergency had arisen or that there was a person in a greater need than themselves:

Sometimes I have to wait to be seen – 2 or 3 hours but you know it's the NHS and there are other patients that have to come before you. If they have come in from A+E. You can wait. Even if you are there promptly. (Basil, 57, Usher).

Yes, the Lady actually came out to me and said "my morning appointment overran, do you mind if I go and grab a sandwich and I was "no, I don't want you to! " (Laughs) And obviously that was fine, everyone has got to eat! And I know they are very committed because they have overran with my appointments before so I know they are very committed to giving the best possible care so you know. So the one time they were late, they told me what was going on so that was fine. (Chris, 31, Norrie's Disease).

The worst we've waited is 3 hours. But of course it's normally where [consultant] has been called into surgery. As we say it has happened to us. We've been that emergency. We will sit. It's fine. [Consultant] always says sorry. But I've said look you've opened on a Saturday for us, I'm sure that you've had better things to be doing or meant to be somewhere else. You can't exactly moan when you've been waiting there for 2 hours – it's fine. I suppose some don't see it like that but you know! (Katie, 31, Stickler).

It eased participants' frustration if they knew why they were waiting or if they were able to go off and do something else whilst they were waiting:

They let me know there was a queue and there were people in front of me. They let me know if somebody cancelled. They let me know straight away. It's things like that they do (Simeon, 46, Usher).

Dave (43, Stickler) gave the example of having had a field test done at 10.30am and still having to wait until nearly 3pm before having had anything to eat or drink. For Dave "a realistic idea of timescales would have been nice", however he was conscious of airing his criticisms too loudly as he recognised "it's incredibly demanding for [consultant]. And I have a great of time and respect for the guy so I wouldn't complain." However Louise was more direct, if the clinic was not going to keep her informed of delays she would go off and attend to her own needs:

No, I just think it is their problem. I am long running... I am 41 I have been going to these appointments since I was a kid so if I am hungry, I just think, you know "I am going for a brew". (Louise, 41, Usher).

Katie (31, Stickler) and Angie (43, Stickler) also appreciated the fact that if they were running late, the clinic staff were understanding and would do their best to make sure that they got seen.

Some participants however made the point that as well as communicating delays it has to be done in such a way that all patients can access it:

Some of them [let you know about delays] but some of them don't. Even those that do, they put it up on the notice board, like [clinic] they put it up on a sign and I just think, how dense are you 'cos you

are in there 'cos you haven't got 20 20 vision, so what is the point of putting up an electronic sign saying you are running 40 minutes late when people in there can't see very well. (Harriet, 38, Stickler).

When your eyes are dilated you can't see anything, no notice or anything. (Holly, 49, Usher).

However at one of the clinics that Diane attends with her son Dean (29, CRS) they will let the patients know if they are running late, take a mobile number and call back when it is time for the appointment. Diane describes this as, "much more patient friendly." Sandra (mother of Max, 11, CHARGE) had suggested a text message service where patients could go off and be called back 20 minutes before their appointment, to her local major hospital. This would be especially useful for her son Max as he finds sitting still very difficult:

He will walk for miles. He doesn't like being enclosed in a certain area, he doesn't play with toys, he just likes to walk. It destresses him. So I could go walking round the hospital and then if someone said oh you're in in 20 minutes can you make your way back. That's been my great suggestion to (major hospital) – can you just do a text service? Cos it's really hard. (Sandra, mother of Max, 11, CHARGE).

Sandra gave the example of being at the local Eye Hospital and being asked to sit down with her son by a nurse, her response was "I can't make him sit down, he'll just destroy the place!", although it is unclear whether Sandra actually said those words to the consultant, or just thought them, nevertheless her frustration is clear.

And often it has been hard just getting there, finding somewhere to park, walking there, and getting in, so by the time you've even got him in the room you're shattered and really you don't need some lippy nurse saying could you just sit there for half an hour. (Sandra, mother of Max, 11, CHARGE).

Sandra went on to say that as well as dealing with the attitudes of hospital staff, other parents can also put added pressure on parents and patients:

It is not just naughty child syndrome there is sensory issues going on – that sort of thing. But other parents can be awful! Horrible! Some are ok but... I think that's harder. Like Max dribbles and some are like ugh that's disgusting, but I'm quite hardened to it. I don't care but I can see some others would be really upset by it. (Sandra, mother of Max, 11, CHARGE).

However at Sandra's local hospital, the community paediatrician is so well known for being late that they allow patients to ring up and find out how long the delay is before setting out from home:

But they'll let you ring up, and I ask how late is she and they'll be she hasn't even turned up yet! So I say what do you reckon, and she says ooh give it another 2 hours and I'll give you a ring if... so they are quite good. She sees a lot of disabled children. You do get your money's worth when you go in. So nobody minds waiting. (Sandra, mother of Max, 11, CHARGE).

This provides us with an example of the sorts of things that clinics could do to provide the best possible experience for patients (and their carers), although one does have to question whether running two hours late is appropriate in any situation.

Becky also made the observation that her son Josh (13, CHARGE) also finds waiting difficult, and identified a need for quiet places where children who have sensory issues can go:

But Josh gets quite distressed. He can get quite distressed and quite aggressive when he is distressed and if he is very anxious he will flip out so I have learnt over the years that sometimes it is easier to just take him away for a while. Just take him somewhere quiet. (Becky, mother of Josh, 13, CHARGE).

Fortunately for those attending the specialised holistic services at Birmingham Children's Hospital and the Queen Elizabeth hospital in Birmingham there was less waiting around. Because there are so many consultants to see, and tests to have done, patients generally do not have to wait:

It's an all day job. You get there for 9 o'clock in the morning and you don't finish until half past six at night, and you see 20 specialists!

You see the genetics, the eye specialist and everything. (Kevin, guardian of Isla, 8, BBS).

Usually I go for my ears test and my heart test, and my lungs test, and my kidneys test. Some people go for everything and some people just see the dietician. I always see the pain lady. I also see the psychologist because a lot of pain is to do with psychological. I get lots of headaches because I get tired easily. (Paul, 49, Alström).

When I get in the room, that's it I go from one room to the other room. As long as there is no one else in there at the time.... I'm not really waiting a lot around no... That's something I like about Birmingham. (Jack, 27, Wolfram).

The clinics also try and cut down on waiting times by seeing siblings together, or at the same time but separately (Samantha, 21, Wolfram; Nour, 10, BBS). However this lack of waiting or 'down time' was difficult for Rob (27, BBS) who said, "It is part of my learning difficulty as well, I find it frustrating, one person after another."

Participants were asked whether they had the opportunity to meet with other people with the same syndrome whilst waiting for their appointment, a number of patients suggested that they would like to do this, for example Jeff (42, Usher) said, "It would be good to have a bit of time to talk to someone different." Meanwhile Jonathan (12, CHARGE) said, "I would **love** to meet someone with CHARGE. Someone who has my syndrome."

However as was also indicated by a number of participants, the clinic waiting area is not the most appropriate time or place to be meeting other people:

I don't know really if that's the most appropriate place 'cos obviously everyone's going to have come from such a different place, it's more better in the local area that you live in. I think in the clinic would be difficult. (Nina, 46, Usher).

Not a lot of conversations goes on between patients and it's not really encouraged and you need to keep the voice level down so the support group networks need to be promoted more I think. (Dave, 43, Stickler).

Angie (43, Stickler) who has no vision, explained how she relies on someone else coming up to her and introducing themselves at clinics. Similarly at accompanied visit (e) it was only because the patient was talking to the researcher that another patient recognised the first patient's voice and was able to come over for a chat. If the researcher and the patient had not been talking the two patients would not have known each other was in the room.

As described above, the clinic lunch – for patients and their families attending specialised holistic services – gave patients the opportunity to meet other people with the same syndrome. For those patients who stayed in a hotel, arranged by the clinic co-ordinators, before a clinic, or for those adults with Alström during the clinic (as the adult Alström clinic last for 2 days), this also gave participants the opportunity in the evenings to socialise:

It's kind of difficult because some people have more treatment than others and they maybe feel tired or something so we don't socialise, but try to socialise in the evening and have a drink together. That's when we most know about each other's problems and that sort of thing. (Paul, 49, Alström).

8.3.3 The waiting room

To understand more about the experiences of hospital and clinic settings for people with rare syndromes and sensory impairments the participants were also asked to comment on the physical environment of the waiting room. Entertainment facilities, lighting and noise, and seating will be discussed in this section.

Some of the children who perhaps had not answered many questions on their experiences at hospitals, instead leaving it to their parents to answer the majority of questions, were able to tell us what they liked or disliked about the waiting rooms. For example Josh (13, CHARGE) said, "I like Mario Kart". (video game). Maisie (6, Stickler) said "There were story books, there were trains, teddies, stuff like that." However Chloe (11, Stickler) and her brother Ollie (6, Stickler) had some recommendations on things that could be changed to improve their hospital experience:

Chloe: [Hospital] has a small bit for kids bits but there is nothing really for older kids. But [a different hospital] has nothing.

Ollie: They should have a room for younger kids, a room for middle kids. I like that. A group for older kids.

Chloe: They should use the TV not use it for dental stuff. Stop giving us numbers for dental stuff that no one is going to use.

Ollie: I think they should put a TV in all the rooms. All the waiting rooms.

Jonathan (12, CHARGE) also made the observation that the hospitals that he attends do not provide things for older children, “They shouldn’t just supply toys and books for younger children... Something suitable for 12 years old.”

Kirsty also made the suggestion that if toys were available that were suitable for her son it would improve the experience for them both:

If they was sufficient enough to cater for a child, children with special needs like Finlay, like he has to have the lights and the sensory toys. Things like that. (Kirsty, mother of Finlay, 14 months).

At clinic (i) the co-ordinator told the researcher that they had recently invested in some iPads that the children could use, however unfortunately on the day of the clinic they were being repaired and the children were not able to use them. The co-ordinator had also been thinking about toys and games that might be suitable for visually impaired children but recognised that they might not be suitable for children with hearing impairments or that they might disturb the other children at the clinic.

Space to play was also important for Roxy and her children, especially when the distance travelled to attend a clinic is taken into consideration:

When you think they have been sat in the car for 3 hours all the way to [hospital] and they have been dragged round the hospital made to sit in this confined area; there is a couple of toys, but they want to run up and down the hallway because this tiny bit of waiting area is not letting them let off steam in any way. (Roxy, 29, Stickler).

Kirsty's son Finlay (14 months, CHARGE) also needed space to play out of his pram and this was not available to him at the clinics that he attended:

...more suitable areas for children and ain't got no muscle tone, like Finlay, he's got no muscle tone. He can't sit up, so he does a lot of lying down. So that would be helpful, just a little area not the whole big space. Just a couple of mats, padded mats for children that can't sit up and things like that. That would be brilliant. Because that would make my life less stressful, he wouldn't get so stressed out and it would be so much easier (Kirsty, mother of Finlay, 14 months, CHARGE).

For Diane's son Dean (29, CRS) access to the internet was very important;

...[internet] is his access, that's his reading a book, that's his listening to the radio, all the things that people do, stuck in a hospital, he can't do, he needs that wifi, he needs his laptop. (Diane, mother of Dean, 29, CRS).

Wifi internet access codes were written in large writing on a wipe board at clinic (vi) and at clinics (i), (ii) and (iii) there were notices on the wall inviting patients to ask for the wifi code, however it relied on someone being able to see those signs or to have someone with them that could see the signs.

A number of adults who were attending ophthalmology appointments and had to have dilation drops, suggested that there's not much that you can do to keep yourself entertained whilst waiting as it is impossible to read or look at a screen (Gordon, 31, Usher; Holly, 49, Usher; Katie 31, Stickler). Basil (57, Usher) however suggested that he always brought along his cassettes to listen to whilst waiting. In one of Jean's (67, Usher) clinics there was a television with subtitles, however she was unlikely to be able to access this having had dilation drops..

The environmental audits took note of the entertainment facilities available in the clinic waiting rooms – note that at clinic (i) the researcher observed the patients waiting in three different waiting areas, and at clinic (iv) two different waiting areas.

Table 7: Environmental audit – Clinic, entertainment facilities

CLINIC	Entertainment facilities
(i)	<p>1) Ride on toys, play kitchen, building bricks, policeman's helmet, fish tank.</p> <p>2) Colouring, projection of images on the floor that can be changed by jumping or walking on them, 2 boxes of cars and building bricks, push button toys. Bubble tube.</p> <p>3) Books, TV with Disney film, no subtitles.</p>
(ii)	<p>Colouring, projection of images on the floor that can be changed by jumping or walking on them, 2 boxes of cars and building bricks, push button toys.</p> <p>Bubble tube – not switched on.</p>
(iii)	<p>The co-ordinator showed the researcher a small sensory area however it appeared that parents would have to make a special request to use this area, and that this area often doubled up as a consulting area.</p> <p>Colouring, projection of images on the floor that can be changed by jumping or walking on them, 2 boxes of cars and building bricks, push button toys.</p> <p>Bubble tube – not switched on.</p>
(iv)	<p>1) TV showing Disney film – no subtitles or sound.</p> <p>2) Small number of books and a noisy abacus.</p>
(v)	Nothing
(vi)	TV with rolling headline news at the bottom of the screen.
(vii)	Nothing.
(viii)	<p>Lots of toys but hectic and noisy</p> <p>Colouring table – print outs with lots of crayons</p>

	Wii with two controllers Disney DVD playing but quite loud – no subtitles Lego table
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Although toys and things to do were available at clinics (i), (ii), (iii), (iv), and (vii) thinking of the needs of the patients (people with rare syndromes and sensory impairments) the fact that play areas were small meant that they quickly became noisy and hectic, which was mentioned as not being suitable for some of our participants. Most of the things available were only suitable for under 10s and the majority for under 6s, there was nothing available for older children. There was no play space in use for people who liked to be on the floor - it is a shame that the sensory room seen in clinic (iii) was not available. Although not all children would be able to read the subtitles on a DVD, by not including them, access for some children who did require them was blocked. For adults, apart from in clinic (vi) where there was a TV with the news headlines there was nothing to pass the time either. Ultimately the entertainment facilities might not be the most immediate priority for those in charge of funding however waiting is often a central part of the patient experience and if there are things that can be done to make it better they need to be highlighted. Ultimately if people are happy and relaxed before their consultation it makes for a much better experience for everyone.

Participants often mentioned that clinics were noisy, hectic places:

...sometimes there are elderly people with eye problems for different clinics and been sitting in the same place and for them it is not great that there are kids running and screaming. So it is not thought out well in that way... (Roxy, 29, Stickler).

A lot of the times it is very packed. That was the one thing I was shocked when I went to [hospital] the first time. I could not believe how many people were there with similar sight impairment. I could not believe it. It was so packed. So, so packed and it ain't a small area. (Simeon, 46, Usher).

I think most of the environments at [hospital] are just too busy. And I think in an ideal world you'd go all those kids that have specialist needs where too many people, and too much distraction and too much light, and too much noise were really detrimental to how they function, that they would have their own clinic which was much quieter. That's an ideal world I suppose. So that they don't have to cope with as much. (Wendy, mother of Luke, 11, CHARGE).

This was also observed in the environmental audit, which as well as looking at noise and space in the clinics, looked at lighting:

Table 8: Environmental audit – Clinic, waiting area noise, lighting, and space

CLINIC	Waiting area noise, lighting, and space
(i)	<p>1) Very hectic so not much space especially with siblings and young children . Large ride-on toys. Average lighting, small window, no blinds.</p> <p>2) Small area so if one person was noisy easy to disrupt everyone. No room for more than 2 pushchairs/wheelchairs.</p> <p>Poor lighting, lack of natural light, but if needed blinds on small window.</p> <p>3) Large area so could probably move if noise in one area was too much.</p> <p>Large window at one end of the room with blinds. But dark furniture and tired looking – so was not bright and airy.</p>
(ii)	<p>Small area so if one person was noisy easy to disrupt everyone. No room for more than 2 pushchairs/wheelchairs.</p> <p>Poor lighting, lack of natural light, but if needed blinds on small window.</p> <p>Radio playing at reception desk</p>
(iii)	<p>Small area so if one person was noisy easy to disrupt everyone. No room for more than 2 pushchairs/wheelchairs.</p> <p>Poor lighting, lack of natural light, but if needed blinds on small window.</p> <p>Radio playing at reception desk</p>
(iv)	<p>1) General background noise but a large area so you could move to a different area if there was a noisy person near you.</p> <p>Not much natural lighting but lots of lights switched on. Good lighting.</p>

	<p>2) Small corridor and could get noisy – difficult to move to another area. Also abacus on the way was a noisy toy to play with.</p> <p>No natural lighting but lots of lights switched on. Good lighting</p>
(v)	<p>Pretty quiet and enough space to move if you needed to.</p> <p>Good, bright lighting with window at one end with anti-glare film.</p>
(vi)	<p>Pretty quiet and enough space to move if you needed to.</p> <p>Bright and light, but no natural light.</p>
(vii)	<p>Pretty quiet and enough space to move if you needed to.</p> <p>Good bright lighting, bright walls, blinds on windows if needed.</p>
(viii)	<p>Very noisy</p> <p>Sheer volume of numbers</p> <p>Lots of children doing things but also TV playing DVDs loudly.</p> <p>Quite dark and crowded/cluttered probably made it seem darker than it was.</p>

Here we can see a variety of practice, some good, and others with factors that could be improved for participants, which contributes to the patient experience, for example space to move away if the clinic was getting busy or noisy. A quiet, calm atmosphere is valuable. Participants did not speak that much about lighting in the waiting room however it was recorded as part of the environmental audits to show the variety of experience and as well as some of the positive things that can be done, such as the availability of blinds and anti-glare film, or good artificial lighting when there is no access to natural light.

Noise and busyness in the waiting room was particularly anxiety provoking as participants were worried that they would miss being called for their appointment:

And I just sit in the chair. I'd be waiting there all day if I was on my own. Sometimes I feel at [hospital] that they don't take consideration for different people. Even though I've got my cane, and they see me with my cane, but it's only the fact that (wife) hears my name being called out. No one's actually come up to me and said "oh you've got to move round to the other section." It's only that (wife) has heard that I know to move to the different section. (Jeff, 42, Usher).

Yes because if you go on your own that is the most nerve wracking part thinking will I actually hear the person calling my name. That really is very important that the person who calls your name is heard. (Jean, 67, Usher).

In accompanied visit (e) the participant would have missed their appointment unless the researcher had been there, and in accompanied visit (d) the participant complained to the researcher that the staff calling the names were too quiet. At accompanied visit (a) however the nurse calling patients spoke very loudly and clearly, and in visits (b) and (c) the calling was appropriate for the patient and volume of background noise.

The majority of people who found listening for their name difficult were people with Usher, because the people with CHARGE had a parent accompanying them to their appointment, and as will be discussed further in the next section, those attending specialised holistic services, and the Stickler condition specific clinic had a clinic co-ordinator who knew the patients and would go up to them and speak to them directly:

They are really good about.... When I go in to say I am here. Even when I am going to [clinic], I still say "if you could make sure I hear you when you call my name"... But I have never had any problems with that, and then I go into the waiting area either [consultant] or [specialist nurse co-ordinator] will come up to me and say "you ready for the appointment, or come on through" or something like that... (Maureen, 47, Stickler).

What was particularly frustrating for participants was having told the staff that they had a sensory impairment, or when they were carrying a symbol of this (i.e. a cane) this was not taken into consideration:

I just think if they understood Usher and how it impacts on hearing and things like that instead of them all walking around going Donna... "Is it me you are shouting?" you know that kind of thing and for them to actually understand what the cane means i.e. the red stripe on my cane. That would be really helpful. (Donna, 46, Usher).

I know when you go to this eye clinic there are all different eye problems. They have to realise the hearing side of it, people who are severely deaf. The struggle to really hear anything. Sometimes they can be calling my name. Luckily I always have someone with me, they can say "Holly, your name is being called." (Holly, 49, Usher).

Basil also made the point that although he could generally hear his name being called he could not see where to move to next and staff did not always appreciate this:

They say [name] and I stand up and they say can you come this way please – you know I've got a stick here what do you want? They don't come over to you and say would you like to hold my arm. And that's what they should be doing! If you've got a white stick you go over to the patient and offer your arm at least! (Basil, 57, Usher).

This was also seen in accompanied clinic visits (d) and (e). In visit (d) the nurse called through the patient, saw them stand up, but did not wait to check that the patient was following, and in visit (e) the member of staff said 'come this way' and it was not until the patient specifically requested guide support that it was offered.

In order to avoid missing their name being called, and knowing which way to go, participants spoke about keeping, or trying to keep, their eyes focused on particular spots in order to know when names were being called:

You would go to this waiting room and it would be a really huge waiting room for everyone who was attending every clinic in that

building and they were all facing different directions but generally not facing the direction where the person who would call your name would come from and because they were working with so many clinics, names were being called constantly. Different nurses would come in and you didn't know which one that would call your name so you had to watch all of them and when you have got Usher syndrome... and you can't see and you are sat there for hours literally. All you could do is just stare and wait for somebody and hopefully catch your [nurse], you can only look at one person at one time and that is why I never went alone. (Linda, 58, Usher).

There's a central area where you have to sit in chairs and there are lots of doors all around the side, so you never know which door someone is going to come out of and call your name. So I never actually hear them calling my name – my husband does. He'll give me a nudge and say "it's you" but I still don't know if it's in front of me or behind me. (Jean, 67, Usher).

Yes, I think the very frustrating thing is not knowing which direction you should be looking in even if I am with someone, even so, you are looking and seeing which room am I going to be in? All these rooms... I wish I knew which room I was going to be in. (Louise, 41, Usher).

As Louise comments, "I wish I knew which room I was going to be in" a simple thing such as telling the patient which room they were to be seen in, could be very positive in terms of promoting the patient experience. Similarly Nina (46, Usher), whilst recognising that it might not be suitable for everyone, suggested that the display board could be used to display the patient's name rather than just calling it. Another consideration to improve the patient experience was about the seating and the positioning of chairs to ensure that people were more easily able to identify where staff were coming from (Jean, 67, Usher).

In relation to seating, a few participants complained that there was not enough seating. For example Becky (mother of Josh, 13, CHARGE) said, "It's noisy, it's busy. They tend to be very full so you just sit where you can really." Helena and Dave also commented:

Everybody who goes to [hospital] has some sort of wheelchair or pushchair or assistance aid for their child, so there's not enough space – not by a long stretch (Helena, mother of Rosie, 4, CHARGE).

Yup, at busy times there can be a lack of seating available because additional people, again a little bit of forethought there in that a lot of patients are brought to the hospital by a parent, carer or family member and they don't really seem to have thought the seating out for that. At very busy times it can be difficult to find a seat, yup. (Dave, 43, Stickler)

This was also seen in accompanied visit (b) when there were not enough seats in the waiting area for patients and accompanying people. In this instance the researcher and the patient's mother stood, allowing the patient the opportunity to sit. This was important as the patient who already had balance difficulties had just had dilation drops administered and needed to sit quietly whilst the drops began to take effect. Also the patient needed face-to-face communication and because the seats were in a fixed row it was easier for their mother to stand facing them. Harriet, speaking as a mother of two children with Stickler (Chloe, 11; Ollie, 6) spoke about how the fixed seating was not appropriate for her children, especially after a long drive to get to the hospital:

I don't like that the seats are all in a row. It takes you 2.5 hours to get [to the clinic] especially if you have kids and you are expected to sit in front of each, and they are grumpy with you if they bang the chairs. They are expected to act like adults and they are not. (Harriet, 38, Stickler).

Fixed seating in corridors, at accompanied visits (a) and (b) also meant that it was difficult for someone who had a sensory impairment and/or mobility difficulties to walk down the corridor areas as naturally people's legs, and then also pushchairs and wheelchairs, were in the way. Simeon (46, Usher) and Jeff (42, Usher) both mentioned a need for seating that has good contrast with the floor. For Jeff this was particularly important after having the dilation drops, as this impaired his vision even more.

With these needs in mind, during the environmental audit of the clinic, seating was taken into consideration. The types of seating were recorded, taking into consideration whether or not there was good contrast with the floor, whether the seats were moveable, whether there was enough seating, if there was seating available for children as well as adults, and any other features. For example, in accompanied clinic visit (e) it was noted in the waiting area for retina photography there were extra-wide seats for those that needed it.

Table 9: Environmental audit – Clinic, seating

CLINIC	Seating
(i)	<p>a) Comfy, washable sofas, good contrast, positioned around the room – not able to modify arrangement.</p> <p>Not enough seating everyone was in the room together.</p> <p>b) Some soft chairs, some hard chairs, moveable, less good contrast as previous areas.</p> <p>Just enough seats for everyone.</p> <p>Smaller chairs for children.</p> <p>c) Individual chairs in various configurations. Not able to be moved but good contrast with floor.</p> <p>Plenty of spare seats</p>
(ii)	<p>Some soft chairs, some hard chairs, moveable, less good contrast as previous areas.</p> <p>Smaller chairs for children.</p> <p>Just enough seats for everyone.</p>
(iii)	<p>Some soft chairs, some hard chairs, moveable, less good contrast as previous areas.</p> <p>Smaller chairs for children.</p> <p>Just enough seats for everyone.</p>
(iv)	<p>a) Comfy, washable sofas, good contrast – not able to modify arrangement but plenty of seats to choose from.</p> <p>b) Row of seats, cinema style pull down, along wall, not able to modify. Well contrasted with floor.</p> <p>Just enough seats for everyone.</p> <p>Difficult to walk down the corridor as seats on either side.</p>

(v)	<p>Fixed position seats and a number of movable chairs.</p> <p>Some with arm rests, some without.</p> <p>Good contrast with floor.</p> <p>Enough chairs for everyone.</p>
(vi)	<p>Fixed position seats.</p> <p>Arm rests.</p> <p>Just enough seats for everyone.</p>
(vii)	<p>Movable chairs and some fixed seating.</p> <p>Some with arm rests.</p> <p>Good contrast with floor.</p> <p>Just enough seats for everyone.</p>

(viii)	<p>Not enough seats</p> <p>Child-sized chairs at colouring desk and lego table</p> <p>Flip down seats (blue good contrast with wall) in passage way (wider than a corridor but like a corridor)</p> <p>Quite busy to walk past especially for someone with mobility difficulties/sensory impairment/who has had eye drops</p> <p>Seats fixed to wall so not able to sit facing each other</p>
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Once again there were differences in provision across the hospitals, and even within clinics. Overall the seats were in good contrast with floor and there were just enough seats for everyone. However in some cases there were not enough seats, and if there were, they were in a fixed position so as to make communication more difficult. Also as mentioned by Becky and Sandra above, if someone required space away from other people that was not possible. There were no places for children (or adults) to lie on the floor, which as Kirsty mentioned was important for her son.

8.3.4 Clinic co-ordinators

At the specialised holistic services at Birmingham Children's Hospital and the QE hospital in Birmingham there are clinic co-ordinators who work in a liaison role, providing a link between the hospital and the patient's lived experience. These co-ordinators, whilst funded by NHS England under the NHS specialised commissioned services programme, were associated with the charities in the UK that are run to support people with the syndromes under investigation (e.g. Alström Syndrome UK, LMBBS (Laurence-Moon-Bardet-Biedl-Society), and Wolfram Syndrome UK). It is part of the funding requirement for specialised services that patients are involved in the planning and delivery of services and clinic co-ordinators are seen as an efficient way of meeting this objective. At the Stickler clinic at Addenbrooke's hospital there is a specialist nurse for the clinic, and although the nurse is not working for

a syndrome specific charity, and as a nurse has a medical role, in practice for the participants there was no difference in the way that the participants experienced this – which was overall, very positively. For example the father of Mohammed (9, BBS) and Nour (10, BBS) said that the two co-ordinators from the BBS society were very helpful and informative and Jack (27, Wolfram) said “The Wolfram Syndrome people are kind, helpful, listening. They do everything.” Similarly Dave (43, Stickler) described the nurse co-ordinator as “an absolute star” and “a real patient champion.”

It was observed during the environmental audits that the co-ordinators would greet the patients (and accompanying family members) by name, sometimes even hugging and kissing them. There appeared to be a genuine connection between co-ordinators and patients and a desire to help to make the appointments go as smoothly as possible. Katie (31, Stickler) for example spoke about how she was hoping to bring her son up to meet the co-ordinator at her next appointment, as the co-ordinator had been interested in Katie’s pregnancy and the birth of her little boy. As part of the environmental audits co-ordinators were observed providing a whole range of different services for patients such as distributing documentation on how to claim back travel costs, providing sighted guide support so patients could attend clinics independently, playing with patients whilst their parents had a discussion with doctors, providing information on benefits and play schemes for children, and welcoming new patients. Patients also appreciated the administrative support that co-ordinators provided, particularly in terms of having a named contact:

(Clinic co-ordinator) is fantastic, normally if I have any issues whatsoever I just email her, so I emailed her to say I wasn’t well and she said she would rearrange the appointment for me so she was able to do that (Maureen, 47, Stickler).

In accompanied visit (a) a family worker from a voluntary organisation was also present at the consultation, along with the consultant, a second consultant, and a student doctor. It is anticipated that this person might be acting in a similar role to the clinic co-ordinators however neither the patient, nor the patient’s father knew what this person’s role was, other than ‘they worked for [voluntary organisation]’. Indeed when asked if

anything had surprised him about the day, the patient's father said, "I didn't know all those people were going to be there." The family worker took an interest in the patient and was pleased to hear that school and family life were going well, however without the opportunity to gain a rapport with the patient and their family, or specifically ask about the patient's needs, by talking in the waiting area before the consultation, or providing follow on contact details, the presence of the is person in the clinic did not provide the co-ordination which was thought of so highly at the specialised holistic services.

8.3.5 Summary

This section has focused on the reception areas, on waiting and on the role of specialist co-ordinators. The key need for face-to-face communication, for staff to take time and look at patients, speak up, and speak to the patient rather than a companion, were emphasised many times. Waiting was difficult, but participants appreciated information about how long they might need to wait, and why, which needed to be in appropriate formats. Waiting would be made easier if consideration was given to individual needs; appropriate activities for children of different ages, TV with subtitles, or wifi. Noisy, busy waiting areas made it hard for to children to wait, and participants worried a lot about not hearing their names called for their appointment. Waiting is not something that clinicians often see, but it is central to the patients' experience of hospitals. Participants appreciated it when family members were seen together, or separately but immediately following each other. Some participants appreciate (or would appreciate) the opportunity to meet other people with their syndrome, but others would prefer this to be through network groups. Specialist holistic services were sometimes able to include a lunch within the day, which enabled patients to meet in a relaxed way. Clinic co-ordinators performed a vital role in the holistic clinics, as a friendly face, in organising appointments, and in helping with logistics (such as claiming travel expenses).

8.4 Consultation

In this fourth section, the consultation stage of the clinic visit will be explored. However it is not limited to a meeting with the consultant *per se*, but includes tests that occur before meeting the consultant and conversations with other doctors, not necessarily the lead consultant. To illustrate this, the table below shows the different people that the patients who were a part of the accompanied clinic visits, saw during their consultation; where known the official role of the member of staff is provided.

Table 10: Accompanied clinic visit and list of consultations

Accompanied clinic visit	Consultations
(a)	1) Patient was weighed and measured 2) Consultation with clinic lead, 2nd consultant, Voluntary Organisation worker, and student doctor.
(b)	1) Vision test with optometrist 2) Administration of dilation eye drops by nurse 3) Consultation (not with clinic lead)
(c)	1) Vision test 2) Administration of dilation eye drops by nurse 3) Consultation with clinic lead 4) Pre-surgery check with junior surgical doctor 5) Pre-op assessment with nurse
(d)	1) Vision test 2) Retinal photography 3) Consultation with registrar

	4) Consultation with clinic lead
(e)	1) Administration of dilation eye drops by nurse 2) Retinal photography 3) Consultation with registrar 4) Consultation with clinic lead

Keeping in mind the things that are particularly important for people with dual sensory impairments this section explores communication, information, and the environment, in order to more fully understand the experience of hospitals and clinic settings for people with rare syndromes and sensory impairments.

8.4.1 Communication

Patients described how communication at the consultation stage varied. In the best examples there were no communication difficulties as Basil and Jean explained:

It's very good. They know I've got Usher syndrome. It's all recorded. It's clear. "Can you hear us?" "Can you hear me?" And they are very professional. Not a problem there. (Basil, 57, Usher).

[Consultant] does sit reasonably close but not too close. And it's quiet. And I could follow what he was saying. And he didn't appear particularly impatient when I couldn't hear. When I said sorry what did you say – you know. He was fine. I had absolutely no trouble with him at all. (Jean, 67, Usher).

The communication needs of the participants were different, and what is appropriate for one person with Usher for example, may not be appropriate for another person with Usher. However what was difficult for participants was, having highlighted to the member of staff, either on that day or on a previous occasion, that they had specific communication requirements this was forgotten or dealt with inappropriately. For example when talking to Marg staff would forget that she was a lip

reader, and Chris needed the doctor to look at him in order that the sounds did not get muffled:

Half the time, when they are talking they are moving their head away and sometimes I can't catch what they say. I need someone with me as I can't follow what is going on. (Marg, 48, Stickler).

The reason why I was finding it hard to hear was that the consultant of all people, was facing away from me looking at his computer...And [wife] said she was really shocked by their whole attitude there. I think they had me down as a bit of a hypochondriac to be honest. (Chris, 31, Norrie's Disease).

As Simeon mentioned it was the fact that he kept having to tell people to speak more loudly that was particularly frustrating:

Some people speak very quietly. If I wasn't lip-reading I would be lost. It would be nice if people just, I know they are not used to it, but raise their voice somewhat higher consistently so you don't have to keep saying to them 'excuse me I can't hear you, could you speak louder'? (Simeon, 46, Usher).

For Kim, the hospital was making an effort to understand the communication needs of all its patients and Kim had been asked to fill in a long document on her daughter's communication needs, however, she had declined to do so as she believed it would be a waste of time as this information would not be referred to by staff:

...they did have this form you had to fill in, it was called... about communication and stuff like that but I wasn't going to fill it in, because I knew damn well that even if you read it you wouldn't really understand Lily because no one is just going to suddenly take her off my hands and actually if I spent two hours filling in the thing, can I be assured that people are going to read it? No. So for me that doesn't work. (Kim, mother of Lily, 7, Infantile Refsum).

Louise also made the point that it can be difficult for patients to have the confidence to speak up about their communication needs:

I always have to tell them and once I tell them they are better. But I always have to be vocal about it. I am a vocal person, so that is ok,

but my sister isn't, she is not a vocal person, she wouldn't speak up. (Louise, 41, Usher).

In Gordon's case the reception staff understood his needs but the doctor dealt with it inappropriately:

...I went to the hospital and I signed in at the check in desk and I told her I'm hard of hearing, and when you're there they just say your name, instead of shouting it out, I sometimes miss the name, so I went to the check in and said can you bear with me I'm hard of hearing, and that was fine she just kept an eye on me, when it was my turn she just came up and got me. But when I actually went to see the doctor he literally sat there and shouted at me! He might have been aware of the eyes but he certainly wasn't aware of how the hearing worked! He didn't need to shout at me – just speak up a little bit! Don't cover your mouth and I will be fine. (Gordon, 34, Usher).

Conversely, in accompanied visit (e) it was only the consultant, and none of the other members of staff, who was observed communicating with the patient appropriately (i.e. at a louder than average volume and by using the patient's name to attract their attention before beginning to talk to them).

Not only is the loudness of communication important, and not covering the mouth when speaking, but making sure that the methods used meet the needs of patients e.g. giving a spoken description of something if a patient is not able to see it, or providing an auditory commentary when completing a task. For example in accompanied visit (c) the doctor explained to the patient that they were writing notes however this did not happen in visit (e) and after some time the patient asked the researcher what was happening. A few patients also spoke of communication difficulties when positioning themselves for the slit lamp, or for retinal photography. The patient has to place their chin and forehead in the correct position and hold on to two handles either side of the machine, and this is done whilst the lights are off which means that lip reading is not always possible (Nina, 46, Usher). Jeff (42, Usher) found this procedure particularly stressful as he did not hear what was being said, and it was only when his wife specifically said to the person doing the

tests that she needed to switch the light on, tell Jeff what he needed to do, then switch the light off and begin the test, that Jeff understood what was happening.

In accompanied visit (e) when the patient was having retinal photography it was observed that the patient's chin and forehead were pulled into position by the person doing the photography. Whilst it might be anticipated that the person taking the photographs might need to touch the patient in order that they were in the correct position, it would be polite to inform the patient e.g. 'I'm just going to put my hand on your left cheek and move you forward' or indeed to touch a shoulder first and indicate that a movement was required. Also in this accompanied visit the same person who was guiding the patient pushed the patient on their shoulders to indicate that they should sit in a specific chair rather than telling the patient where the chair was and describing the chair to the patient, (e.g. the chair is directly in front of you, there are arm rests on either side and it has a tall back) or guiding the patient's hand to the back of the chair. In this particular instance the patient, as they were not aware what had happened, laughed and apologised for not getting it right.

As mentioned above often when a patient is with another person the hospital staff will talk to that person instead of the patient:

When I was first diagnosed I didn't really notice the difference, when the doctors stopped talking to me all the time, but I'm getting more confident as well in saying, and all the doctors know me quite well. If I see a different doctor, I really feel the difference, they don't talk to me at all. Sometimes the doctors ignore me – I just sit there! (Rachel, 40, NF 2).

100% every time, if you say you have got a hearing loss they stop talking to you and they talk to the person that you are with. Always. You have to go "hello, what was that?" (Louise, 41, Usher).

Louise went on to highlight that in the same way that a family member should not be used when translating foreign languages, a member of the family should not be used when a patient has a sensory impairment:

You are completely left out of the loop and I am “what’s going on, what is happening, why are we coming in here” and [husband] is translating the whole time. And it makes me laugh because they are always saying in the letters “a family member can’t translate for you” and I know they are talking about people [for whom] English is a foreign language and they are saying for your own best interests you can’t have your brother translating for you as they might not get it right but I think, what do they think [husband] is doing? He is translating for me! (Louise, 41, Usher).

If a professional communication worker is used they need to be appropriately trained for working in a medical setting. Angie for example, described her pre-op blood test as “pretty traumatic” as her communicator guide

...wasn’t telling me what was happening and all that was happening was someone tapping my arm. Someone was tapping my arm but no one was actually telling me what was happening. (Angie, 43, Stickler).

Knowledge of the patient and also some medical knowledge is useful as Rachel (40, NF 2) explained. Previously the hospital had supplied Rachel with a BSL interpreter however she now goes with her own support worker who drives Rachel to her appointments, knows how best to communicate with her, and has learnt something about Rachel’s medical history and NF 2:

It’s better to have somebody who goes regularly because they start to know, because before the hospital would book and every time it was someone different. So they had no idea about my background or about NF 2, so it was really hard for them to sign it. It wasn’t really fair to throw them in that situation. Now I have someone who goes, who knows what they are signing. (Rachel, 40, NF 2).

The hospital had also provided Vera with a BSL interpreter for her appointments, however Vera uses hands-on BSL and the interpreter was not able to communicate with Vera, this was despite Vera’s daughter specifically asking for a hands-on BSL interpreter:

The second time I had an interpreter but they weren't suitable for deafblind. So we were struggling a bit with the communication. I was talking about allergies but I wasn't able to spell it and the interpreter didn't understand. It wasn't the right interpreter. It made me feel stupid because of the problems with the communication. (Vera, 83, Usher).

The third time this happened Vera's daughter had to take over the communication which left her daughter feeling 'shocked' and 'awkward'. This returns us to Louise's point of the appropriateness of family members providing communication support and also when things are asked for and they are not acted upon it is frustrating. However some parents whose children did not use formal communication methods, or needed support in doing so, spoke of the importance of including them as parents and communication partners in the patient's care:

...for someone like Dean where you've got a communication issue particularly and a learning disability,... that the person with the most day to day knowledge and the most communication with that patient is not just tolerated but actively involved in all decision making and all interaction, because that a) makes their life easier and it makes the patient's life easier, and therefore makes their job quicker... (Diane, mother of Dean, 29, CRS).

Sometimes their understanding of disabled children is really poor. The eye surgeon was going [makes baby noise)], come on, a) he's 11, and b) he can't hear you. Whatever you need to do just do it! Or tell me and I will communicate it to him. (Sandra, mother of Max, 11, CHARGE).

Sue and Kevin (guardians of Isla, 8, BBS) also suggested that if possible doctors should learn some basic BSL or Makaton in order to "come down to [the child's] level" (Sue, guardian of Isla, 8, BBS). In this way medical staff would have a greater understanding, of how children who use such communication methods were feeling.

Finally, one particular difficulty that a number of participants faced in relation to communication was that of 'accents':

Yes that's a problem with foreign doctors. I can't understand them and they can't understand me. So it's a bit of a waste of time in a way. I need an interpreter! (Doug, 60, BBS).

Participants who struggled with accents were often embarrassed to say so or felt awkward in raising the issue with the researcher:

They are friendly staff and you can understand a word they are saying as well. They speak English unlike some places – whoops... (Laughs). (Jack, 27, Wolfram).

Firstly [consultant], he's not English, it's not his first language and he talks very fast and this is the problem with deaf people and they do have problems with foreigners. I have to be careful sometimes because I get frustrated and say things to myself and occasionally they come out. You have to be careful not to offend people these days. (Paul, 49, Alström).

For example Jeff's (42, Usher) wife was keen to point out that the staff were 'lovely' but that her husband struggled with non-English accents. However as Jean mentioned it is not just difficulties with 'foreign' accents that patients might face:

They are ok but the (local) accent can be hard to understand and to lip read so sometimes I might turn to my husband and say what did they say? (Jean, 67, Usher).

Although a person might not be able to do anything about their accent, an awareness of what contributes to making an accent more easily understood, e.g. facing the person directly, not covering the mouth, not standing directly in front of a light source, speaking at a moderate tempo, and writing things down if appropriate would help to facilitate improved communication in a hospital and clinic setting. An alternative or additional step might be a communication protocol that asks if a person is communicating with a patient appropriately.

8.4.2 Information

The consultation centres around the exchange of information between patient and doctor. For all patients there is a need for clear, accessible

information, and generally the participants in this piece of research received this:

...they speak to us in a way that we can understand. I do think it is a really good clinic the way it is all set out. The way they treat us. They don't speak to us like we are children or something. They speak to us in a way that we can understand. (Jessica, 21, Wolfram).

All the doctors are really good. They explain everything. It is a really tiring day but it is good. They see all the doctors. (Muna, mother of Deema, 14, Aalia, 12, Lilas, 9, BBS).

And they answered [my questions] to me in a language I would understand that wasn't all like Doctors' language so they were saying it how I would understand it and I understood it. (Jack, 27, Wolfram).

Participants were however aware that clinicians might not always be able to answer their questions or provide more detail due to the lack of information on the syndrome due to its rarity (Maria, 55, Wolfram; Paul, 49, Alström). However this can still be frustrating or confusing for participants:

...that's just the way it is sometimes. They try to explain as much but it's never enough to my mind. I like lots of detail – I've got that sort of mind. (Doug, 60, BBS).

It is important that clinical staff pitch their explanations at the right level and in most cases staff appeared to do this:

[Doctor] talked to your level. Didn't talk down to you, didn't talk up to you. If you asked him to explain something to you he would do it in your language. If you wanted the technical stuff he would give that to you as well. (Paul, 49, Alström).

In accompanied visit (e) the patient was talking with the consultant about recent advances in stem cell therapy and the consultant checked the patient's understanding of 'retinal pigment epithelium' before continuing. When the patient responded that they did know, the consultant replied "I knew you would!" and they both laughed. Indeed it was nice example of

a positive patient-doctor interaction which is hoped would be a feature of all consultations.

However some participants did provide examples of when they left the consultation without the appropriate information:

No I'm left with a million questions. No the doctor... when you go to the Eye Hospital they don't tell you anything. You're left... you go in they take the photos... especially the one in [former city], it was a case of you turn up, they do what they want to do, and they say right off you go. See you in a year. They didn't tell you any information. (Gordon, 34, Usher).

...I had 3 procedures carried out simultaneously on my eye by [consultant], but I really was confused about what was being done. I wasn't given anything in writing, it was just verbally explained and it was all sort of in medical terms. (Dave, 43, Stickler).

However the participants in this research project, due to their sensory impairments, as well as the very nature of having a complex, rare syndrome, had particular requirements that other patients might not need, for example the chance to touch and hold equipment, the opportunity for 'time out' during the course of the consultation, access to both written and spoken/signed information, and information in a range of formats e.g. large print, electronically, or in symbols. People with sensory impairments often have a greater need for context to understand what is being said. They may need an outline of what will happen beforehand as they do not get the sensory anticipatory clues. Or patients may need vocabulary explained as they may not be familiar with certain terminology, even that which other patients may have heard. Patients may need breaks to account for fatigue, as they are working harder than other patients to listen, and above all going to a clinic appointment is stressful in and of itself. Furthermore some of the patients spoke about fatigue as one of the 'symptoms' of their syndrome (Paul, 49, Alström ; Jessica, 21, Wolfram; Phillip, 28, Wolfram).

One example is that some participants might prefer to use touch to gather information, and clinical staff need to make sure that this is available. For example when Jonathan (12, CHARGE) had a blood test he was able to look at the equipment, as well as exploring the hydraulics

system on the bed. When Chris was exploring the possibility of cochlear implants he was able to touch the different processors that were available:

So there was no uncomfortableness cos sometimes you have to say “can I actually touch it” there was none of that. They put it in my hand straight away basically so that was fine, so yes there have been plenty of opportunities to handle equipment. (Chris, 31, Norrie’s Disease).

Participants also found it useful to have verbal information followed up with written information supporting what has been said:

Yes I prefer to read it for myself. I read the one about the different Usher types 1, 2 and 3. But I prefer to read it myself after someone has talked to me. I feel as though I have got everything right then. (Vera, 83, Usher).

Instead of them just trying to say what they have said I prefer to have a letter, with what they have said, and how the tests have gone, and what the verdict is. So if I have to try and explain it to (support worker) or someone else, if I’ve got the proof there I can pass it on to the people that need to know. Instead of trying to explain, or getting (wife) to explain what they have said. There’s communication breakdown sometimes. (Jeff, 42, Usher).

If there are a lot of doctors all talking it’s hard to get everything. But they will email me afterwards which is brilliant. (Rachel, 40, NF 2).

Having written information and details of their medical condition was important to participants as they needed this information for professionals working in other fields e.g. education or social care, and to ensure other service needs were met (Sandra, mother of Max, 11, CHARGE; Diane, mother of Dean, 29, CRS; Holly, 49, Usher). Louise needed information about her vision for her application for Personal Independence Payment (PIP), the new benefit replacing Disability Living Allowance (DLA) and was shocked when the doctor knew nothing about these benefits:

[Doctor] knew nothing about DLA and he didn’t even know what PIP was and I just thought, you’ve got no awareness about what it is like

to be a visual impaired person in the world and yet this is your job, and these are the things you should know. It might be medical things but you should know what it is like to live as a visual impaired person and all the things that.... So I just left there and thought, that was absolutely pointless. A waste of time. (Louise, 41, Usher).

An awareness of the challenges for people managing sensory impairments, along with a rare syndrome, and that these go beyond the medical context, is important.

Furthermore, when information is provided it needs to be in the appropriate format according to the patient's needs, for example relating to font, large print, paper colour, via email, etc. Samantha and Jessica (21, Wolfram) explain their frustrations in not being able to access information independently:

Samantha: We always have to get mum to help us. It's a bit frustrating knowing I can't sit down and read it myself. That I have to rely on mum.

Jessica: Exactly. It would be nice just to be able to get something that I can read myself rather than going oh mum can you read this out.

Similarly Angie (43, Stickler) was disappointed that the only information available to her from her rheumatologist was in print, which she could not access:

The rheumatologist gave me a book on back pain and I couldn't read it! It's basically here you are, here's written information and go away and read. (Angie, 43, Stickler).

Angie also spoke of how she had found that the Occupational Therapist struggled to adapt the advice and information that they usually gave to sighted and hearing patients to meet Angie's needs:

...even though they knew about my sight and hearing loss because we had talked about it at the pre-op at great length, it seemed to throw them if you know what I mean. They didn't seem to know how to adapt their spiel, what they normally say to patients, to me. (Angie, 43, Stickler).

Likewise Jeff (42, Usher) who needs printed materials in large print and on yellow paper, received his pre-op information on white paper and in small print. However Chris gave the example that although he was unable to see his audiogram results, they were described to him, thus making them accessible:

I am just thinking I don't know, how for example you would portray an audiogram in an accessible format so from that point of view, no but it isn't a criticism, it is more that I don't think it is possible. The important thing is, and they didn't do this in [previous hospital] is they explain the results to me. So I understand them. So from that point of view, that makes it accessible. (Chris, 31, Norrie's Disease).

Due to the fact that many patients with rare syndromes have a complex medical history and are seen by many doctors, this information needs to be shared appropriately between different departments and hospitals. Becky (mother of Josh, 13, CHARGE) provided an example relating to her son's surgery. The cardiac team should have spoken to the cleft repair team, however this did not happen and it could have led to complications for Josh. Wendy (mother of Luke, 11, CHARGE) and Sandra (mother of Max, 11, CHARGE) did however give examples of medical staff working together to ensure that their sons had to have the fewest anaesthetics possible, or that bloods were taken or teeth were cleaned, when the patient was having another procedure in order to minimise the distress for their child, however Wendy's example shows the work that she has to do as a parent in order for this to happen:

His bloods, for instance, they'll print off the forms and give them to me and then it's a case of going into the hospital and when the anaesthetist comes round saying there you are, this is what we need. And then saying to the nurse, you're definitely going to get that blood, because I feel the need to do that so I know that it's all going to be co-ordinated because I don't want him coming out and needing a load of bloods done and it hasn't happened. So we put off things like blood tests until he is having a general anaesthetic in order to try and minimise disruption and confusion for him. (Wendy, mother of Luke, 11, CHARGE).

Kevin and Sue (guardians of Isla, 8, BBS) spoke about the difficulties that they had since moving house, and that information about Isla was not shared between hospitals. This meant that Isla was going to have to have various tests repeated which was not only invasive, frustrating and time consuming, it was also costing the NHS a lot of money.

Sue: They will not transfer [Isla's medical records] up here for her. So it means we have to do everything from scratch again.

Kevin: Because when they request anything they have to wait a minimum of 12 weeks which is not very good with regards to them. Like they were going to waste thousands of pounds doing the sleep study from scratch again to get the results. It's not electronic yet. If it was electronic they would be able to access it from anywhere in the country.

Some of the young people also mentioned that it was important to be honest with them regarding procedures. For example when talking about cannulas Josh (13, CHARGE) said, "They say they are not hurting but they are." To which Josh's mother Becky followed up with,

It doesn't help when they say it's not going to hurt and then it does hurt. It would have been better if they were more honest about it I think.

Similarly Chloe (11) and Ollie (6) felt that that the medical staff did not appreciate how uncomfortable having dilation drops was for patients, just saying 'they sting a bit'. Likewise in accompanied visit (b) the patient (a child) had to have dilation drops. The patient was anxious about this whilst waiting and the researcher observed the patient having their drops done, which required restraint by two people. Afterwards the patient's mother commented how she did not like that nurse as 'her attitude was not very good' and seemed to 'not have a very good way about her'. Sandra also made the point that children with sensory impairments do also feel pain and need to be treated with respect:

I think sometimes they treat deafblind children as if they are lumps of meat, who need stuff done on, forgetting that they can feel pain, or maybe they just want to get it done. I think instead of one size fits

all, they should realise that every child is different. (Sandra, mother of Max, 11, CHARGE).

As well as patients receiving information from medical professionals, the consultation should also provide the opportunity for patients to ask their own questions and give their opinions. As with many patients attending clinics, participants in this research project spoke about coming with a list of questions, making notes, and writing things down. This was observed in accompanied visit (a) when the patient's father came with a small notebook and noted down things that the consultant said, as well as referring back to it when asking his own questions.

One of the participants did however make the observation that if there is only one specialist in the country, and there are difficulties between the patient and the consultant, or the patient wants a second opinion, there is nowhere for the patient to go:

We were told to think about it, take some time and [we] wanted some more information about it and every time we tried to question [consultant] he got very rude, very defensive, angry, defensive, you could hear it in his voice. He didn't want to actually discuss it with us. (Participant).

However particularly for those patients attending specialised holistic services, or condition-specific clinics the opportunity to speak to someone who knows about their condition was very important. Participants spoke about being treated as an equal and having the opportunity to tell their story:

But for years a lot of the doctors I saw didn't have a clue about it and when I was trying to explain it to them they were like "you haven't got the degree in it; you don't know what you are talking about. (Phillip, 28, Wolfram).

In clinic visit (a) the patient's father said that the best thing about the clinic visit was the opportunity to ask all the questions that he had, and for the patient it was to be able to tell the doctor some of the changes that he had noticed because of the medication and that "I want to keep on with the [medication]." Being listened to and having the opportunity to speak is very important for all patients, however parents and carers of

some of the young people we spoke to highlighted how they are the key person in their child's care and this must not be forgotten, and their knowledge and understanding of both the patient and their syndrome is equal to that of the medical profession. When thinking of improvements to the patient experience Diane commented that it was important that :

...the person with the most day to day knowledge and the most communication with that patient is not just tolerated but actively involved in all decision making and all interaction, because that a) makes their life easier and it makes the patient's life easier, and therefore makes their job quicker so that would be the point. (Diane, mother of Dean, 29, CRS).

8.4.3 Environment

As with the other areas of the hospital, an accessible environment during the consultation is important. Noise and lighting was noted during the five accompanied clinic visits:

Table 11: Environmental audit – Clinic, consultation room noise and lighting

CLINIC	Consultation room noise and lighting
(a)	<p>1) Bright, light, airy room – but no natural light</p> <p>Door to corridor was kept open</p> <p>2) Bright, light, airy room – but no natural light.</p> <p>No noise coming through from the corridor</p>
(b)	<p>1) Bright, airy room.</p> <p>No noise from the corridor outside.</p> <p>2) Very bright room.</p> <p>Lots of clutter around the room.</p> <p>Patient was making a lot of noise so unable to note if there was noise from outside.</p> <p>3) Bright, airy room.</p>

	Noise from corridor outside
(c)	All five rooms were light, bright and airy, with no noise from the waiting areas disturbing the consultation.
(d)	<p>1) Bright light room.</p> <p>Door to corridor was kept closed.</p> <p>2) Dark room as photographs being taken.</p> <p>Noise from machines and only a curtain (not a door) to divide the room.</p> <p>3) Lights were dimmed to examine the eyes</p> <p>Curtain dividing spaces. Not private and voices could be overheard. Noisy building works going on around.</p>
(e)	<p>1) Lights were dimmed to take photos</p> <p>The room was shared with another patient who was having different type of photographs taken</p> <p>Noisy machines</p> <p>2) Lights were dimmed to examine the eyes</p> <p>Curtain dividing spaces. Not private and voices could be overheard.</p>

In all cases the basic lighting was appropriate to the patient's needs, although of course when lights were turned off patients would not be able to lip read or get clues from the environment. It was only in clinic (b) that the consultant gave the patient a warning that the lights were going to be turned up again. Noise however was particularly problematic. In clinic (b) although the door to the corridor was shut, the noise of children playing and babies crying could be heard inside the consulting room. The patient's mother mentioned this to the researcher as one of the reasons why crowded busy clinics are not suitable for her son. At clinic visits (d) and (e) noise was a particular problem. The consulting areas and retinal photography areas did not have walls on four sides and a

door - they opened on to larger areas and had dividing curtains. In clinic visit (e) another person was having photography at the same time as the patient, the conversation between the other patient and the person doing the photography was distracting, as was the whirring of the machines, and also meant that there was little privacy for the patients – each could hear what was being said to the other patient. In clinic visit (d) the noise of building work during the consultation meant that the patient said “I am really struggling with background noise” and asked if they could go to another room. The patient was told that there was nowhere else to go, which in the post-clinic interview the patient said really surprised them. They were pleased however that the consultant would be providing them with all that had been discussed in a written format, as they felt they had missed quite a bit of the conversation.

The open plan nature of some of the clinics was also slightly confusing to some of the participants that we interviewed. For example Jeff and Nina commented:

[Consultant] doesn't take me anywhere private. He just says it where he is. There's one person doing it with one person, and another doing it with another person. I've never really gone into a room at [hospital]. (Jeff, 42, Usher).

Obviously they have the instruments there that they are going to use so it is a bit cluttered. But I think it's ok, the only thing about it is that you're in one bit and there'll be another person being seen in another bit so it's a bit open plan so sometimes they might be talking to you and they might go and speak to the consultant about this and they'll go off and they'll try and find the consultant or whatever. So possibly for some people they might say there's not enough privacy. (Nina, 46, Usher).

A few parents spoke about the desire to have toys or something to play with in the consulting room as it is quite difficult to keep a child entertained as well as listening to what the doctor has to say in an often stressful situation. Kim (mother of Lily, 7, Infantile Refsum) commented, “the intensity of it all is beyond anybody's complete and utter... you know.” Sally also said:

...it would actually be quite helpful if in the consulting rooms they had more toys and things for children to do, because you do have that discussion time with the professionals about your child's treatment and issues and children do get quite bored at that point. I think it would be helpful to have a few more toys in the consulting rooms, as well as the waiting rooms. Obviously they do in the child development centre but in the other hospitals they don't so much (Sally, mother of Poppy, 5, CHARGE).

Kevin (guardian of Isla, 8, BBS) welcomed the fact that at the specialised holistic services there is often someone there to entertain Isla whilst he speaks to the doctor.

8.4.4 Summary

In this section we discussed the consultation; with an emphasis on communication and information. Participants experienced different levels of communication, with some clinical staff being very aware of individual needs, whilst others were not. Even when participants had been asked about communication needs, they did not always believe that staff would refer to this information. Unfortunately, poor communication skills were seen and described far more often than good ones. Participants who could not see, or whose sight was very impaired, were not told what was happening during appointments, for instance, why lights were going off, or why nothing was 'happening' when doctors were writing notes. Participants were frustrated by staff talking to their companions rather than them and by inadequate or inconsistent provision of communication support (where needed).

Most participants felt they received clear information, but as their conditions were complex, so was the information, and as they were rare, there was not always much information available. Participants needed information which sighted/hearing patients did not, such as being able to touch equipment and they needed information in appropriate text formats (large print, braille, digital). As listening is hard for people who are hearing impaired, they needed information backed up later, by text, such as emailed reports on the clinic visits which they could read in their own time, in their own format or have read to them. Such report information also needed to be used for supporting benefit claims and

similar procedures. Participants also felt strongly that staff should be honest about procedures which hurt and should understand better the patients' perspective on procedures which were uncomfortable or inconvenient, such as retinal dilation. Clinics were usually well lit, but sometimes unnecessarily noisy (because doors were left open or some areas were only bounded by curtains). As hearing impaired patients need louder speech, they were also uncomfortable about non-private areas for conversation.

8.5 Beyond the clinic

For a large number of patients there was relief that their appointment was over and they were keen to get on and make the (often long) journey back home. Sometimes there were changes, for example an alteration in medication or a referral to a different consultant, or as was seen in accompanied visit (c) an emergency operation was needed. Often however patients would simply go on their way and either book the next appointment or wait for it to come in the post, as identified in the first section 'Appointments', taking us full circle in documenting the process of attending a clinic. Furthermore for those patients who attended many appointments it was just the end of one appointment before the next one started.

Many participants did however identify an ongoing need for information, support, and advice in relation to their or their family member's medical condition. This information, support, and advice was identified in terms of medical knowledge and research, emotional support, practical guidance on how to manage various aspects such as tube feeding, as well as just meeting and being with other people with the same syndrome.

Participants did however draw distinctions in the support and guidance that they received (or wanted to receive) from hospitals and that which was provided by charities and other organisations. These two areas of provision will be analysed in the following sections.

8.5.1 Support and follow up from the hospital

Participants identified a need for support and follow up from the hospital in two ways; the first was as a point of contact if things changed between scheduled appointments, or if patients had any particular worries or concerns. The second was at the time of diagnosis or when a significant change happened e.g. certification of sight impairment.

At some of the clinics there was a clear, identified plan of what to do if there were any problems between clinic visits, whereas in others patients were left floundering. Gordon was frustrated that he had no-one

to contact if there were changes in his vision and when asked to suggest any overall improvements to his clinic experience said:

Just the way of getting in touch. If there are any changes I would like to be able to phone the doctor and say... or even just leave a message with the receptionist, 'I've got this problem, can you help?' (Gordon, 34, Usher).

Another example of this lack of identified pathways for support away from the clinic setting related to Jeff (42, Usher). Jeff had lost his glasses, which had been prescribed at a specialist hospital clinic, and he did not know what to do about it before his scheduled appointment in January (this was October); consequently he was going to go without his glasses for 4 months.

Conversely however, Sally (mother of Poppy, 5, CHARGE) and Ellen (mother of Maisie, 6, Stickler) had both been given the contact details of their daughter's ophthalmology consultant with the instructions to get in touch if there were any concerns. Also as was seen in section 8.3 above, those patients who attended clinics where there was a clinic co-ordinator had a central point of contact with whom they could be in touch if any problems arose. For example, Maureen (47, Stickler) had been told, every time she left the clinic, "you know if you have any problems, to contact [clinic co-ordinator], or to contact our office or whatever."

Other participants suggested what they might do if they felt that they needed to get in touch with someone – usually this was contact the local GP or the community paediatrician. However Lily's (7, Infantile Refsum) mother spoke about emailing the consultants if she had any questions or worries and Phillip (28, Wolfram) identified his local endocrinologist as the person who he would get in touch with if he was having any difficulties and then this person would contact the specialised holistic service if necessary.

Participants identified wanting support, information, and advice from the hospital or consultant at the moment of diagnosis, and often felt this was lacking:

No, no I didn't get no, nothing, no guidance as to what I do next. All they said, all I could remember them saying is they would write to

the GP and the opticians that made the original referral. And that was it. (Linda, 58, Usher).

...I was like ok, I'll deal with whatever comes along, but no one was giving any information about if he had it, and what you could do and what, this, that or anything. I was left to get on with it myself actually. (Kirsty, mother of Finlay, 14 months, CHARGE).

I thought when we got the diagnoses with Sticklers, I thought [clinic] should have given us a sheet really about it. And the support group - they should have really told us about it so we had somewhere to go. (Harriet, 38, Stickler).

However as Louise (41, Usher) mentioned, it is not until a person is 'in the right place' that support and information networks can be tapped into. Helena described her initial thoughts and emotions regarding her daughter's diagnosis of CHARGE:

It's hard to say it was all a bit of a blur. The geneticist spent quite a bit of time talking to us. Talking it through. We could only take in about 10% of what someone was saying. (Helena, mother of Rosie, 4, CHARGE).

Similarly Simeon, speaking of the overwhelming nature of information on first diagnosis said:

...when you are not familiar about the information it's easy to disregard it because it just seems like a lot of jargon people are talking about not realising that the very thing they are talking about is pivotal to your wellbeing, survival and living a better life. (Simeon, 46, Usher).

In the accompanied clinic visits, there were no new diagnoses made, although in the case of visit (c) the patient did have to have emergency surgery, yet in all consultations the patient (or their parent) was asked if they had any further questions, and the patient's (or parent's) understanding of this was double checked by the doctor. However on leaving the consultation, and in a brief discussion with the researcher afterwards, the father of the patient at clinic visit (a), as well as the patients from visits (d) and (e), said that they were unsure of a particular aspect of the information that they had been told. This suggests that

even if patients are given information, in stressful, complex situations such as consultations, this information might not always be remembered, and if it is, not correctly.

Even if hospitals had just referred patients on to other organisations or charities that would have been useful as Maureen explained:

...it would have nice for the clinic to have given some supportive information such as Sense or Deafblind UK or some type of support group even right down to telephone numbers for the RNIB or the RNID; anything would have been nice. They are fantastic: very supportive but I think as far as outside support now you have been diagnosed this is who you can reach out to, cos that's the one thing I have had real hard problems with. (Maureen, 47, Stickler).

Although some participants had been diagnosed a number of years ago, for example Doug (60, BBS), Nina (46, Usher), and Jean (67, Usher), those feelings of disbelief and bewilderment, and the lack of appropriate support and guidance from the hospital, stayed with them and affected their overall patient experience.

There were specific times when participants identified the need for explicit support or guidance from the clinic, for example Alice wished that support had been in place when she had lost the vision in one eye:

I think if it was more personal and they offered like guidance and counselling and support and stuff because that's probably one thing that I would have benefited from. I think losing an eye at the age of 10 is a big deal but to them it was 'oh you've got another eye' – so it's ok, it's not a big deal. But I don't really see it that way. (Alice, 17, Stickler).

Simeon (46, Usher) also identified a general need for counselling for issues surrounding his dual sensory impairments. Basil (57, Usher) was however fortunate to receive hearing therapy and counselling at one of the hospitals that he attends, however he was concerned that the funding for this could be cut at any moment. Similarly, Katie (31, Stickler) was very appreciative of the information and support provided to her when she was thinking about and under-going CVS (Chorionic Villus

Sampling) testing in order to see whether her unborn baby would have Stickler.

8.5.2 Support from charities and organisations

Since hospitals and clinics were perceived, in general, as not providing ongoing support to patients, participants generally sought support and guidance from charities and organisations associated with specific syndromes and/or sensory impairments. Participants spoke about getting support from a range of charities and organisations including Sense, Deafblind UK, RNIB, Guide Dogs, Action for Blind people, RP Fighting Blindness, NDCS (National Deaf Children's Society), HSI UK (Hearing and Sight Impaired UK), local visual impairment charities, as well as charities relating to specific syndromes – Alström Syndrome UK, CHARGE Family Support Group, LMBBS (Laurence-Moon-Bardet-Biedl-Society), Stickler Syndrome Support Group, and Wolfram Syndrome UK. This support came in the form of information packs and leaflets, annual conferences, fun days, and Facebook groups. Sometimes professionals in the field would provide information and advice while at other times it was people with a rare syndrome, or their family members, who supported other family members or people with the syndrome. Participants often dipped in and out of these charities and organisations as was appropriate to their needs.

Participants were appreciative of the information that organisations provided, whilst recognising that there is not a 'one-size fits all' approach to information and support. Dave (43, Stickler) had spent a lot of time thinking about the support needs of people with Stickler and his observation was that the needs of people with a rare syndrome (in this case Stickler), vary considerably as does the presentation of their syndrome,

So the impact of the syndrome is very very personal to every single individual which makes it [a] very difficult condition to support properly, both from a clinical and social side because it is something you have to live with to understand the impact of and because it affects every patient more or less uniquely. (Dave, 43, Stickler).

Dave, as well as other participants such as Simeon (46, Usher), Becky (mother of Josh, 13, CHARGE) and Nina (46, Usher), recognised that

leaflets were very useful way of obtaining information about a particular syndrome. Sally (mother of Poppy, 5, CHARGE) commented, “I think that orange pack [about CHARGE syndrome] that Sense developed is great. I saw it online and I have actually got a copy of it as well.” Kevin (guardian of Isla, 8, BBS) had printed various leaflets on BBS from the LMBBS website and had included those in Isla’s hospital passport in order to provide information for the medical staff caring for Isla.

Information in the form of leaflets was not the only form of support that participants were looking for – meeting others with the same syndrome was very important:

I would love to meet someone with CHARGE. Someone who has my syndrome. In CHARGE syndrome someone doesn’t always have the same symptoms as someone else with CHARGE. To some degree we understand what it’s like. (Jonathan, 12, CHARGE).

I would [like to meet other people with BBS] cos then they are all the same. [My daughters] think that they are the only ones. (Muna, mother of Deema, 14, Aalia, 12, Lilas, 9, BBS).

It would be nice to have a social group in this area. That would be nice. I would love a social group where I could meet others with the condition or others with deafblindness, but there is no one I know round here that has deafblindness. (Jack, 27, Wolfram).

Jean (67) and Holly (49) were part of an Usher mentoring scheme. This is something that Linda was hoping to be involved in:

But it would be nice to have a one-to-one with someone that had that same experience. You know the difficulties you do have coping. There’s a lot to cope with, the deafness and the eyes. (Linda, 58, Usher).

Linda’s experience perhaps indicates that services are only available in certain areas or that they are not always advertised in the best way possible.

Phillip had been in touch with other people with Wolfram at conferences and via the internet and he had found this very beneficial:

...I feel they understand more and that's like, now I have friends around the UK with WS, we do tend to speak often now, and that has helped me a lot. (Phillip, 28, Wolfram).

However there were no organisations in Phillip's local area for people with visual impairments so he was setting up his own support group.

For parents just to be with others in the same situation was very useful as Helena explained:

Going to the CHARGE conference and that Sparkles (specialist toddler group for children with multi-sensory impairments) group it felt like for the first time that we belonged. They were tube fed, they had balance problems – we were just amongst peers as opposed [to] being the odd ones out. To feel as though you belong there, and have supportive peers that's amazing. (Helena, mother of Rosie, 4, CHARGE).

Unfortunately for Kirsty this was something that she was missing at the moment:

If there was support... ...more information, a place where I could go and meet other children with CHARGE and I ain't got to sit in a room and go on he's got CHARGE he got this, this, this and this. I want someone else to go oh my child has got CHARGE and I am right 'and this is Finlay', we haven't got to explain ourselves to each other because we are both in the same position. (Kirsty, mother of Finlay, 14 months, CHARGE).

Becky (mother of Josh, 13, CHARGE) and Wendy (mother of Luke, 11, CHARGE) also mentioned the Sense Siblings Days which gave their sons' siblings the opportunity to meet with other children who have a brother or sister with a multi-sensory impairment.

The syndrome specific support groups - Alström Syndrome UK, the CHARGE Family Support Group, LMBBS and Wolfram Syndrome UK – all organised a yearly conference in which people with the syndrome, along with family members, medical professionals, and associated

professionals such as teachers and equipment suppliers could come together and share experiences and specialist knowledge surrounding the syndromes. Jack (27, Wolfram) commented, “They have opened my world up.” When asked about the BBS conference Rob (27, BBS) said, “I enjoyed that.” Becky (mother of Josh, 13, CHARGE) described what she got out of the events, “Emotional support and moral support definitely – from the CHARGE Family Support Group and they run family days where we can meet other families and that kind of thing.” Kevin and Sue (guardians of Isla, 8, BBS) described the importance of the BBS conference to them:

Kevin – If it’s the first time they go to a conference it can blow their minds.

Sue – You’re in awe of everything and everybody. The first conference we went to years ago I felt in awe of everybody – I didn’t know about BBS. I’d never dealt with it in all my life. I found out from BBS that you get blindness and that. And I was overwhelmed. I learnt such a lot. And talked to some of the other parents there, and it made me realise I am not on my own.

Kevin – We learn something new every time we go down.

Some parents of children with CHARGE also mentioned that coming together with other parents on line was very useful way to share practical tips and hints on managing the complex medical needs of their children, which are not explicitly shared by medical professionals:

I think that the Facebook group is useful, things like posting semi-medical questions, really funny little things like I keep waking up and find that my child’s gastrostomy has come undone and they are covered in milk and every parent of a child that has had an overnight feed knows what’s that like, and for someone to say, oh there’s this lock that you can get, which no-one particularly says to you, it’s all these little things, which don’t register on professionals’ worlds because they have lots of other things to think about but when you are the person who has got a bed constantly being drenched in milk it is your priority, so you’ll go to the Facebook page now and people will put those things up. It’s a short cut to what

people have historically had to learn the hard way. (Wendy, mother of Luke, 11, CHARGE).

You often go to medical appointments and you get told what the problems are and what the solutions are but it's actually you come away, and it's the parent's job how to manage life. And nobody tells you that in an appointment. So you are handed a child who has a Mic-Key button and a tube, this where you get your syringes from, this is the milk you have to use, this is how you do it but it takes a parent to say "when you go out it's really helpful to have a bag to put the milk and the tubes in and if you take a 10ml tube out is a lot easier to manage than a 50ml tube. And it's helpful to take sterilised water with you because it's hard to find." It's that day to day management and all those little things to make life easier that you never find out in an appointment and it's other people who make life easier... Those people have helped us as a family more than any appointment. (Helena, mother of Rosie, 4, CHARGE).

The fact that Helena finishes with "Those people have helped us as a family more than any appointment" suggests that the needs of people with rare syndromes and their families are not being fully met by medical professionals. It is good for everyone that parents posting on Facebook are able to help with this, but it would seem appropriate that there are other ways, allied with specialist clinics, in which they can find this information.

Specialised support from Sense was identified as being particularly useful as the specific nature of dual sensory impairment was recognised as Becky and Sandra explain:

CHARGE Family Support Group we are in touch with. That's it apart from Sense. We are very involved with Sense but to be honest that is personal choice. I mean there is so many support groups out there for the individual bits of Josh's condition, but I think when you have got a child with complex needs I always find it difficult to go to a group with visually impaired kids...because you also have the hearing impairment and the other stuff so out of personal choice we are just involved in the two. (Becky, mother of Josh, 13, CHARGE).

We get support mainly from Sense. We did go with [specialist charity] but they are really not geared up for children with additional needs, they say that they are but they're not, - they've pulled actually from doing stuff with children with additional needs. I've moaned for years that they are only a charity for children that are deaf and everything else is fine. I think that they went into that because a lot of parents did complain about it but they realised it was just too big a bag. (Sandra, mother of Max, 11, CHARGE).

Sense's specialist knowledge about dual sensory impairment had been useful in supporting participants to get the appropriate educational and social care assessments (Poppy, 5, CHARGE; Josh, 14, CHARGE; Dean, 29, CRS). Megan's (18, CHARGE) mother however identified that the scope of Sense's provision was dwindling, which she was frustrated by, as she felt she had nowhere else to go, and Sandra identified that although excellent advice was available, there was not much practical support that Sense could provide:

I'm not involved in any other charity apart from Sense. They are very good. But again there's no money. They've all left! So they don't really do anything any more because they don't have any money. But the advice is fantastic. (Sandra, mother of Max, 11, CHARGE).

Some participants did however acknowledge a 'proceed with caution' when connecting with other people with the same syndrome, either online or face-to-face. For example Dave (43, Stickler) and the parents of Maisie (6, Stickler) spoke about the difference in the North American and British medical attitude and perspective to Stickler, and that people might be given erroneous medical information:

...it's a bit of a dangerous minefield with the Facebook groups because there's a lot of issues and rumours that get around that which are not necessarily challenged. (Dave, 43, Stickler).

Similarly for Jean, although she enjoyed meeting other people with Usher, she suggested that one must be prepared for some negative aspects as well as positive ones:

Meeting all these people with Usher has the potential for actually not being empowering; it's disempowering you because you become aware of the support you might need – a guiding arm, a helping hand, lip reading, and people doing sign language and you kind of get drawn into this deafblind world, which you've never really thought about yourself in before and you really don't want to think about yourself as being the sum total of your eyes and ears – you have to be a bit careful that you don't get into that trap. (Jean, 67, Usher).

Dave makes the important point that support and information has to be relevant to the individual in terms of their personal characteristics and individual needs:

I do feel that there needs to be a lot more peer support – role models for people to follow and that's another comment. That's reflecting different experiences of gender, the age ranges, that kind of stuff, so there needs to be a reference point. (Dave, 43, Stickler).

For example Dave, along with Jean (67, Usher) and Vera (83, Usher), spoke of their needs at this moment in time, particularly in terms of 'getting older':

I think because as I have got older, the Stickler's issues have got more progressive so where as I have managed to get by all the time in the past, they are becoming more of an issue now and I need to start managing that so that is an interesting point. (Dave, 43, Stickler).

Similarly Vera was feeling more isolated as the small number of people with Usher that she knew had all passed away. This serves to remind us of the particular needs of older people with non-age related dual sensory impairments.

8.5.3 Summary

This section examined what happens beyond the clinic appointment, what support participants needed from both the hospital and from other organisations. Where something had changed, or been decided, at a

clinic (for example being certified as sight impaired) they felt they needed support, and this was often lacking. Pathways for support after the clinic were unclear for some, but where there were clinic co-ordinators, participants did know who to contact. Referrals to support organisations such as Sense would have been welcomed by many. These organisations did indeed support many participants with dealing with the impact of their conditions. Many participants said they would like to be in touch with others with the same syndrome, and where they were, they found this useful.

8.6 Additional points

During the course of their interviews participants often spoke about their experiences with hospitals, medical treatment, and the health service in general. Staying overnight and GP Services will be discussed in this section.

8.6.1 Staying overnight

A number of participants told us of their experiences of overnight stays in hospital, either related to their rare syndrome or as a result of unrelated health problems, and unfortunately in a lot of cases the experiences were quite negative. Basil (57, Usher) commented, “There’s a whole load of problems associated with [overnight stays] and it’s extremely stressful. We come out feeling worse than when you went in.” Participants gave examples of not being able to get to the toilet, not knowing where their meals were, not being able to hear the doctors, and being denied their hearing aids. For Jeff (42, Usher) he was unable to get the specialist inpatient detox treatment that he needed as none of the facilities in England felt that they were able to support his needs as a person with dual sensory impairments.

Jean and Basil spoke of their experiences of overnight stays:

It wasn’t a very good experience because I couldn’t always follow the consultant when he came around with his little minions. When they came round and talked about the patient in front of them... ..It wasn’t very good at all. I think at night time the lights went low and you couldn’t see what was going on very well. (Jean, 67, Usher).

You have to keep on ringing the buzzer explaining that I am blind, that I can’t go to the loo on my own. And I have to wait ages and reluctantly they do it. They’re so short of staff. It’s not fair. It’s alright having one staff for X number of patients but if you’ve got a deafblind person there, that person needs more help. They can’t be equal to other patients who have got their sight and hearing. (Basil, 57, Usher).

Jean also spoke about how tiresome it was to keep informing the nurses of her sensory impairments as they changed shifts. Basil also remarked

that it was important that all members of staff, not just doctors and nurses must be aware of his dual sensory impairment.

As has been mentioned in previous sections a holistic approach to patient care often promotes the patient experience, however things have to be thought through carefully. For example Angie (43, Stickler) appreciated that there was the connection between the ophthalmology and rheumatology departments at Addenbrooke's hospital and she was pleased that her knee operation was taking place at Addenbrooke's however this meant that she had no one locally to pop in and visit her as her family were at home a couple of hours away.

As shown in other sections of the report there were times when hospitals were able to provide the support that patients needed, even when it went a little beyond the usual practice. For example Chris (31, Norrie's Disease), on two occasions, was given a private room so that his wife was able to stay and provide communication support. This does however raise the question of whether a family member should be providing communication support and what would have happened if Chris' wife had not been able to accompany him.

Basil was keen to make suggestions as to how to improve the experience for people with dual sensory impairments whilst on a hospital ward:

If you're an inpatient we should be given an option of a sign above our bed, an eye sign with a line through it, and an ear sign, so that they know you are deaf, so that they know you are blind and wear hearing aids. It's optional. And hearing aids should be next to the bed, so they know that I can't hear. And they need to explain the procedures when my hearing aids are on, before, during and after. Which they don't. They move things around. (Basil, 57, Usher).

Basil went on to suggest that the experiences of patients with dual sensory impairments need to be addressed by charities and the NHS:

And [the needs of deafblind people are] overlooked and Sense and DB UK really ought to get in touch with NHS England and explain that this is just not fair. It's not right. They must address the needs

of people who are vulnerable and have dual sensory loss. (Basil, 57, Usher)

In agreement with Basil's suggestion we would suggest the experiences of people with dual sensory impairments, as inpatients in hospitals, is an area in need of further analysis.

For parents staying with their children in hospital it was an extremely difficult time as Helena (mother of Rosie, 4, CHARGE) explained, "Absolutely horrendous. Awful. Thankfully we haven't been in recently but it's always traumatic. It's possibly more traumatic for us." Likewise Kim (mother of Lily, 7, Infantile Refsum) said, "Hospital stays are the most hardest, most exhausting and stressful things."

Parents spoke about being unable to leave their children's side:

I have to stay with her. They haven't got the facilities to take care of children on their own... ..and I sleep by the side of the bed and what happens is someone comes in and relieves me. Because they don't feed you. And if you want a shower you have to wait for someone to come in and relieve you. There's no privacy. (Sue, guardian of Isla, 8, BBS).

So yes I will always go with Lily, I will always be there for her if she wakes in the night that's up to me. That's a great difficulty because now you know, I can't leave her for 5 minutes so I don't get cups of coffee, I don't get cups of tea, I can't leave her because there is no one to help me. You can't leave her with someone she doesn't know. My only saving grace is if I have a toy she will play with for 10 or 15 minutes. I can get someone to watch her quickly if I am lucky so I can get a cup of tea. (Kim, mother of Lily, 7, Infantile Refsum).

Jen (mother of Megan, 18, CHARGE) found that the cleanliness of the parents' bathrooms left a lot to be desired, "It was disgusting!" Jen also mentioned, along with Dean's (29, CRS) mum Diane, that the noise of all the machines and lights meant that it was difficult to sleep anyway.

Something that made the experience more bearable for Helena was a private room:

Hospitals are just horrible places. The only time when it was more bearable was the surgery when she was 7 months old for her gastrostomy and Mic-Key button insertion and we had a private room – she was the only one in it – it was huge and there was space for us to be in it without feeling as though we were in the same bed. And it made the world of difference to have that space for ourselves and to have a light, well lit room. The rooms at [other hospital] are what I imagine it's like for a battery chicken! Unbearable! (Helena, mother of Rosie, 4, CHARGE).

Wendy spoke about the work that she did with her son Luke (11, CHARGE) prior to medical procedures to enable him to understand and make sense of what was happening. Wendy was happy to produce symbol booklets for her son, but nevertheless it involved a lot of extra work on her part, particularly when she is caring for a child with complex needs and two other children.

As she had transitioned from children's to adult services, Alice (17, Stickler) for the first time had to stay alone in the hospital. This was not a pleasant experience for her as the following quotation shows:

I had a lot of operations before and my mum was always able to stay with me. Last summer I had one, and I was 16 then, so I had to stay on my own then for 2 nights and it was terrifying! (Alice, 17, Stickler).

When Diane's adult son Dean (29, CRS) is admitted to hospital she always accompanies him (sleeping in a chair by the side of the bed) whether it is ITU (Intensive Therapy Unit) or a general Men's Ward in order to support his communication and personal care needs. She leaves the medical team to get on with their role but at all times provides support to Dean:

I mean generally, we're just sort of part of the team. Right, you do the nursing, I'll do the rest of it, and I'll make sure that what's chosen for him for a meal, is something that he will, a) will eat, and b) can eat and c) I'll make sure that he eats it, they haven't time to do that. (Diane, mother of Dean, 29, CRS).

Diane then went on to speak about the importance of being able to trust the nurses so that she would be able to go off and have a short nap, confident that they would come and get her if Dean needed anything. For Sandra (mother of Max, 11, CHARGE) it appeared that the nurses relied on her too much, phoning her up at home to ask her how to change a feeding tube or a tracheostomy. Sandra found this very worrying. Now however as her son has got older and with increasingly complex needs the hospital does not have an appropriate bed for Max to stay in overnight. If he needs a procedure he is first on the list and the hope is that he will not need to stay overnight. Understandably Sandra was concerned as to what would happen if there was an emergency and Max did have to stay overnight:

They don't have a bed for him at [major hospital] so he couldn't stay overnight. So I don't know what they would do... So they tend to put him first on the list, not just because he's more vulnerable but because they don't know what they'll do with him. He tends to get seen first, get done, and then get sent home because he'll need a bed with full wooden cot sides. Oh we've got a cot, but he's too big for a cot. He will escape from anything! Because he needs humidified air or oxygen at night they can't risk him pootling off down the corridor at night time. They just say ooh we don't know what to do. But at one time someone's actually going to have to give it some thought. They said would you be able to lie next to him and I said yes, but I wouldn't be able to prevent him from climbing over me! (Sandra, mother of Max, 11, CHARGE).

8.6.2 GP services

When participants mentioned difficulties with their local GP services the problems centred around a lack of awareness of the communication and mobility needs of patients with sensory impairments, as well as exasperation that when requirements were identified they were forgotten about or not acted upon at the next meeting. Participants also preferred to see the same GP as it helped to build trust between the patient and the doctor but it also meant that they did not have to keep on explaining

about their communication requirements or describing their syndrome. Thirdly, although participants recognised that GPs do not know everything about all syndromes, they appreciated the efforts of anyone who was open to learning a little bit about their syndrome.

As Jack (27, Wolfram) was no longer able to email his GP he was unable to contact the GP independently, describing the situation as “a bit of a nightmare.” Jeff (42, Usher) spoke about his frustration that as he needs communication support at the GP surgery, but the times of appointments do not fit with the hours that his support worker is available –so his wife has to accompany him, otherwise it would be a month’s wait for an appointment. This once again raises the question as to whether family members should be providing communication support in medical settings. In one example reported by Louise (41, Usher) her GP would only provide a telephone consultation. Louise cannot access conversations on the phone, so the GP proceeded to call Louise’s husband and conduct a conversation with him about Louise’s needs. This represents practice which did not meet her needs either as a patient or a person. This angered Louise so much that she put in a formal complaint and changed her GP. Maureen describes her frustration with the local GP surgery as they seem unable to remember to communicate appropriately with her:

I can tell them 100 times that I’m hearing impaired and they will still put their hands on their mouth when they are talking and never looking at me while they are talking so I mean I always find it a struggle when I go locally because I never get to see the exact same Doctor, and even if I see the same Doctor they don’t remember me from Adam so, but every time I will say, ‘I am hearing impaired and can you look at me and can you repeat that again.’ (Maureen, 47, Stickler).

For Jean (67, Usher) and Basil (57, Usher) the environment of the GP surgery is challenging as Jean describes:

Accessing the GP surgery is awful. There is a ramp up one side and steps. The steps are ok, I can do steps, but then you are confronted with this sliding door which apparently slides open, but it’s so dark when you are looking into the interior it’s really bad. So then you go

through one set of doors, and there's the reception and it's a short walk from the door way to the reception and the lighting is just awful, and it's all very grey and quite dark, and it's dark lighting. (Jean, 67, Usher).

However in Basil's case because he cannot see the scrolling display board that calls through the next patient, the doctor comes to meet him and takes him in the lift to the consulting room. Likewise the receptionists guide him to the door or to a seat to help him avoid tripping over children running about. The situation for Sandra (mother of Max, 11, CHARGE) was also quite positive and she would have a telephone call with her GP first to see if it was necessary to bring Max for an appointment. As Sandra describes it "we've built up quite a good relationship. I think they trust me there." Basil and Sandra's examples demonstrate some of the simple things that those involved in providing health services can do to support the needs of people with sensory impairments and/or complex needs when they attend hospitals and clinics.

The relationship that Sandra has built up with her GP is important to her and helps promote the best outcome for her son. Although it is not known for sure, it is anticipated that this relationship has developed over time and is based upon accumulative knowledge on both sides. A number of participants expressed a desire to see the same GP in order to build up this shared knowledge about themselves and their medical condition, as Marg and Rachel explain:

I prefer to stay with one doctor rather having different doctors all the time, then you have to keep telling them everything over and over again. (Marg, 48, Stickler).

Well my GP, they always give you a different doctor. They have stupid booking system and every time you go it is someone different. I tried to always see the same lady, she was very good, but then she retired. And now there is one who is really good but she is only part-time, so it is very hard to get an appointment. They don't know anything about NF 2 or they tell me a load of rubbish. (Rachel, 40, NF 2).

Unfortunately some participants felt that their GPs were not interested in learning more about their or their child's medical conditions as Jean and Kevin explain:

I said "Do you want me to explain about Usher?" and she said "yes but let me get on with what I am doing" – so she doesn't. She doesn't want to know. She wasn't interested really. Which was disappointing. (Jean, 67, Usher).

We've had one medical practice where the doctor has said yeah she's got [BBS], I don't want to learn about it, because he says she'll probably be the only one he'll see in his lifetime, so he's not prepared to waste time and learn about it. (Kevin, guardian of Isla, 8, BBS).

Kevin (guardian of Isla, 8, BBS) was however very pleased that at their new surgery "[they] are quite open to finding out about her, about BBS, actually study it; we've had some thinking about actually going to the conference." Kevin, Jean and other participants recognised that doctors might not have the time or inclination to learn in detail about all possible syndromes but a passing knowledge would be a good place to start as Jen and Kirsty explained:

You can take Megan to the doctors and say oh yeah she's got CHARGE syndrome and they say 'oh what's that then.' The medical team have NO understanding of CHARGE. They're like what's that then. You're like 'Google it'. It does get you quite bitter because you go to the doctor with a problem and they don't know what's wrong with your child anyway. (Jen, mother of Megan, 18, CHARGE).

I had to explain to my GP cos they had never even heard of it. And the health visitor was like "CHARGE, what's that?" In the end it was like, "look it up on the internet!" (Kirsty, mother of Finlay, 14 months, CHARGE).

8.6.3 Summary

This section discussed aspects of NHS care not necessarily linked to clinics; staying in hospital and GP clinics. Hospital stays were often very difficult for patients with sensory impairments, with hospital staff not understanding their communication and mobility needs. Patients often

had to spell them out again and again. For children with rare syndromes and their parents, hospital stays were very stressful, and they had to work hard to help their children cope. In visiting GP surgeries, participants raised again the issues of inaccessible information; and not being able to get consultations in appropriate formats. Where this was working well, they had built up relationships by seeing the same GP over some time.

9 General themes

9.1 Mobility, communication, and information

9.1.1 Mobility (and orientation)

The patients had difficulties with mobility in their journeys to and through clinics. In many cases, their difficulties were the same as those of anyone visiting hospitals – they found car parking difficult, and their journeys took a long time. Of course some of the patients were wheelchair users or had other physical difficulties in walking, but so are many others visiting hospitals. They also had difficulty finding their way around complex sites, where there are few clear routes and labels and signage are inconsistent. In this respect their difficulties were similar to others but significantly exacerbated because they could not see signage that others could see, and they could not ask their way as easily as other people, because they were not necessarily able to see the landmarks others would point out, or because they could not hear the answers clearly. As well as this however, they were different, in that issues which seemed probably simple to most other patients, could cause significant anxiety. One of these for example was difficulty in managing toilet facilities because they could not identify toilets, did not know where the flush was, and so on. One of the patients mentioned a simple strategy, a leaflet for patients explaining the colour coding of toilets, which could also include where the flush was, where the signage about men's and women's toilets was and perhaps include information about the type of soap dispenser and drying arrangements. This could be explicitly read to patients who could not read print but also be available in large print. While it is unlikely that all the toilets in a large hospital will conform to one pattern, in one place or one clinic, they might.

9.1.2 Communication

The patients had difficulties with communication in their interactions with staff when attending clinics. Like other patients who did not have sensory impairments, they found that reception staff were often very busy trying to multi-task and so clear communication could be difficult.

They found waiting times stressful, like many patients, and wished for clearer information. Their difficulties with this were similar to those without sensory impairments but significantly exacerbated because they could not pick up so easily on incidental information about how patients were moving through clinics by hearing patients' names called (they had enough difficulty with their own) or seeing patients move about. There was often little that they could do in waiting rooms because they could not see or hear TV, and there were no materials in large print. For some of them they were also attending with a whole family, all of whom needed to see clinicians. Unlike other people, even apparently simple things like being called for an appointment, could cause them serious problems – they could not take their eyes off the receptionist waiting to lip read their name being called, or strained to hear over the noise of a radio. Unlike other people they found that staff spoke to their companions, instead of them, if they had difficulty in hearing. It is unlikely of course, that most staff are *unwilling* to try to help. However, they may not know how to help, or how to identify people who might need help. A simple question protocol, a discussion of communication needs, could be implemented *across* an organisation. It could be the simple question; is this communication OK for you? As a simple question it could of course have multiple answers, some of which are very complex. However, for the vast majority the simple answer would be 'yes' and even for those for whom it is 'no' the adjustments required would be fairly straightforward – please speak louder, can you read me the form. For the one in 100, or probably fewer, who need something more complex, obviously staff might need to seek further advice.

Remembering to ask the question is an issue of training, but it is not necessarily complex training. Thus while reception staff may not be highly paid and highly trained staff, they could be compared to staff at checkouts who are trained to ask 'do you have your loyalty card' – and do so at every interaction, without overly annoying customers, so reception staff (but not only them) could be trained to ask about communication needs. For the most part, the answers will be simple and could be dealt with – and as a further step they could offer some simple alternatives; Shall I speak louder? Can I read you that form? This would be entirely within the spirit of the Accessible Information

Standard which needs to be implemented across the NHS by July 2016 (Sense et al no date).

9.1.3 Information

The patients had difficulty with getting information throughout their interaction with the hospital, from making appointments to all the clinical situations and beyond. There are complex systems involved in almost all NHS arrangements and most patients find that they have difficulty navigating appointment systems and they do not always leave hospital with the information they wanted or understand exactly what has been said. However, the participants' difficulties with information were made worse because they could not necessarily see all the available information, read the posters or directions, hear the loudspeaker advice about how delayed appointments were. They could not necessarily read the leaflets that they were given. But their experience was different from other people's because they could not pick up environmental and incidental information about how the clinic would run; in which room they would see who, where those rooms were, and so on. Although they might greet the clinical staff who met them, they might not be able to see the name badges they wore, telling them who they were interacting with, or they might not hear the names properly. While it does require some organisation in advance, a written programme of who they were going to see, in which room, with what intention, would be a relatively simple, but very helpful device for ensuring that patients were well informed and that they could anticipate what was going to happen. Not being clear about what will happen causes considerable anxiety and therefore can make all the other issues with which they were grappling, (seeing, hearing, communicating and getting about) as well as dealing with clinical issues, even more difficult. Once again this would be in line with the Accessible Information Standard, as the document could be printed in different formats, read or signed to patients if they needed it, and even if things changed, they could then be communicated clearly.

9.2 Connectivity, multiplicity, rarity, and individuality

When issues relating to mobility (and orientation), communication, and access to information are intertwined with the complexities of a rare

syndrome four themes which describe the patient experience emerge: connectivity, multiplicity, rarity, and individuality.

9.2.1 **Connectivity**

Connectivity, by which we mean the linking together of the various aspects of a patient's being, for example communication requirements, mobility needs, means to access information, syndrome specific issues, medical symptoms, and care needs, as well as home life, personal relations, work or education commitments, is the first overall theme that is drawn out from the patients' experiences. This connection links patients to clinicians, as well as clinicians to other clinicians, and patients to other patients.

Specialised holistic services went some way to providing this connectivity, by bringing together specialists focusing on a specific rare syndrome as Caroline and Rachel explain:

It is a very very nice team. I see the ENT surgeon and I see them since I was diagnosed. What I like in [hospital] is the continuity. I always see the same people every single time. You don't have to have to keep telling them each time. (Caroline, 45, NF 2).

It's a specialised clinic for people just with NF 2. It's brilliant. It's a really specialist clinic for NF 2. The doctors just focus on NF 2. They all work together and talk about it. (Rachel, 40, NF 2).

Participants also spoke about these clinics 'getting things done' whereas other local hospitals were slow to act:

It definitely speeds things up. When the doctors here get a letter from them it moves things on. It felt like [local hospital] were not taking as much notice as they should have been. Nour's ankles were really weak and could not get the supports for her, insoles, but we went to [specialised holistic service] and mentioned it to them, and then the next week we got an appointment and everything was ok! (Imran, father of Mohammed, 9, and Nour, 10, BBS).

Nevertheless patients often still need to attend numerous appointments at their 'local' hospitals. For example, Phillip (28, Wolfram) as well as attending a yearly specialised holistic service attends three other 'local'

hospitals relating to endocrinology, ophthalmology, diabetes, and his bladder.

Similarly for patients attending condition specific clinics, often only one aspect of their syndrome was being dealt with, as Dave and Alexa explained:

I would like to actually go to a clinic where I am supported for the whole syndrome. (Dave, 43, Stickler).

All the associated problems like the joints and things like that were not really addressed because [consultant] was an eye specialist and so we didn't receive that and we didn't get offered any help... We all suffer from like painful joints so that would have been one of the things would have been really helpful would have been some information about where we could have got some help with that. (Alexa, 27, Stickler).

Likewise some of those attending general clinics, where connectivity was not so strong and the understanding of the whole experience of what it is like to be a patient was weaker, suggested that a combined approach to learning about the patient and the syndrome would be beneficial:

...if they could do a bit more joined up thinking that would be really helpful. For kids and for families and for them actually cos I think they miss out on a lot of information between clinics that they don't pass on. (Becky, mother of Josh, 13, CHARGE).

We were quite interested in the combined clinics that they are trying to do in Birmingham, cos that sounds like a really good idea because we have found things quite piecemeal, and if there was a co-ordinated approach that would be brilliant. I don't know if for us whether it would be worth it going to Birmingham for that but if there was something at [local hospital] that would be fantastic, because it means fewer appointments – you see everyone in one day. Everyone has an appreciation of the whole picture of the CHARGE child. Which would be really good. (Amanda, mother of Jonathan, 12, CHARGE).

However for some patients when the reality of their communication, mobility, and access to information needs are taken into consideration a single clinic approach to a syndrome might not be appropriate:

Actually I think it would be too much to have the two at the same time. Actually take back what I said, it would be too much. 'Cos then I would have been prodded in my eyes and my ears and then that would be just too much. (Nina, 46, Usher).

Nevertheless the sharing of results, and understanding of the person's needs would remain crucial, should exist, even if various aspects of the syndrome are covered on different days:

Often at specialised holistic services, when the patient was seeing the same people, communication was easier, as was mobility as patients got to know the building. However the opportunity to see the same consultant, whenever possible, was very important:

I would say that really helps to see the same person and not have to talk about history. It is not easy to talk about history and it is a waste of space and it is just a waste of their time. (Kim, mother of Lily, 7, Infantile Refsum Disease).

Getting to any appointment, whether at a specialised holistic service, a condition-specific clinic, or a general clinic requires a lot of time and effort from patients and their families, in terms of time off school and work, arranging childcare, long distances to travel and the associated costs. Whilst this is true of all patients, as has been demonstrated above, this is particularly true of patients with sensory impairments and clinicians need to be aware of this. One way to lessen this load would be if schools could deliver certain things such as vision screening, or if the telephone consultation as identified by Helena (mother of Rosie, 4, CHARGE) could be adopted more widely.

Furthermore clinicians need to be aware that a patient's sensory impairment and/or rare syndrome impacts upon various aspects of their lives and not just in a medical sense, for example patients may need medical documents to make other things work, for example in relation to claiming benefits, social care support and education:

...whether it's in health or whether it's in social care or whether it was in education, which of course, that's another bugbear of mine, but it's a long... it's pulling ... it's trying to get all those aspects pulled together and looked at as one person. (Diane, mother of Dean, 29, CRS).

Clinic co-ordinators, as seen at the specialised holistic services, and the Stickler condition-specific clinic, occupy a particularly useful role in acting as central liaison point for patients. They provide patients with a pivotal point of contact with the organisation and running of the clinics in terms of managing appointments and also in emergencies. Clinic co-ordinators also provide patients with links to other means of support, information and advice. Other voluntary organisations could get involved in similar ways, to provide support for people with rare syndromes, learning from the clinic co-ordinators what were the most useful things for them to do.

Participants spoke of wanting to connect with other people with the same syndrome, and when people had these connections they generally spoke highly of them:

I feel [other people with Wolfram] understand more and that's like, now I have friends around the UK with Wolfram, we do tend to speak often now, and that has helped me a lot. (Phillip, 29, Wolfram).

I have spoken to a few people and some people I have just observed or read about them and it has given me the courage to just press on and try and find help, find my own niche and my own way to deal with my circumstances in my own way. (Simeon, 46, Usher).

The opportunity to meet other patients is important, and although it might not be appropriate for everyone, if hospitals could arrange a space for patients to meet and help with introductions this could be very beneficial, as people with sensory impairments may not know there are other people in the same room with the same syndrome, or are not always able to employ strategies such as catching someone's eye, or engaging in small talk unprompted.

9.2.2 Multiplicity

Being a patient with a rare syndrome that causes sensory impairments, by its very definition involves dealing with the multiplicity of experience on a daily basis, from managing appointments, to care needs, knowledge and expertise of the syndrome and communication methods, to medications and treatments:

He has growth hormone therapy daily so he has injections daily at bedtime. (Becky, mother of Josh, 13, CHARGE).

You can't look back and regret these things but managing her care when she has been ill has been really hard work... And taking time off for her surgery. And it has taken its toll on our marriage, a lot of the care fell on my husband at the time because he's got his own business, so in theory his time is more flexible but his business suffered, my parents dropped what they were doing and have come down here, it's always a struggle and continues to be so. (Helena, mother of Rosie, 4, CHARGE).

I'm hungry every minute of the day. I've had enough of diets having been on 1,000 calories for years. I've just had enough – it's too much for me. I've been a bit more relaxed. I've been trying over the last year to lose some weight – I've lost a couple of stone. (Doug, 60, BBS).

However clinicians do not always recognise this multiplicity of experience as Diane explained:

[Dean] is not a cardiac patient one day, and an ophthalmic patient another day and a whatever, he's all of them, 24/7, all year! They are just concurrent. (Diane, mother of Dean, 29, CRS).

Patients are 'in the round', they are not just one bit of the body at a time, and they have to deal with this, even though clinicians don't. Clinicians can treat each bit at a time but patients cannot play this 'game'. They MUST be seen in the round and all needs met as Dave and Wendy commented:

[Consultants] don't necessarily see the full impact of Sticklers on you and your life, so one particular area might be clinically not serious enough for them to act on as a single issue but as a combination of issues, sight loss, hearing loss, mobility; it can paralyse you for quite a long time. It is something the consultants need to be more aware on the clinical side. (Dave, 43, Stickler).

No I think they understand complexity but they don't understand CHARGE. And I think that's a way to summarise I think, a lot of the clinicians that we come into contact with understand complexity but they just don't understand the MSI complexity to it. They'll understand in terms of complexity and how many medical appointments you have got, but they don't really comprehend for example with Luke, how fatigue fits into all of that, and how his processing takes much longer, so any eye tests, hearing tests that you do with him it just takes so much longer and they don't necessarily have the specialism to make that environment better for him. (Wendy, mother of Luke, 11, CHARGE).

Participants were aware that clinicians were often involved in research and were keen to ask questions or hear about the latest developments. It often provided participants with a sense of 'I must be in the best place' as Nina explained:

And the good thing as well is that you feel as though they have your interests at heart. They say "Oh we're doing this research to try and find this genetic cure", or also what is quite good is that the consultant will come and see me, and the other guy will go and get the consultant and they will remember me from the year before and say oh yes this is your condition. And one time, the year before, he said oh yes your DNA has gone to Denmark and it is working very hard over there – and I said oh I would rather be in Denmark myself (laughs)! It's good in that respect – you feel as though you are in the right place for it. (Nina, 46, Usher).

Only three participants (from 52) spoke of feeling as though they were specifically being seen as research 'guinea pigs' which frustrated them, and they felt that they could do nothing about it as to remove themselves from that consultant might put their health in jeopardy.

One of the consequences of the ‘multiplicity’ of having a rare syndrome that causes sensory impairments is the conflicting information that a patient may receive; for example Alice (17, Stickler) saw audiologists at two different hospitals one of whom said she had a hearing loss, and the other said that she did not. Angie (43, Stickler) gave the example of the orthopaedic consultant suggesting that she attends the gym, however because of her visual impairment she is unable to attend.

One practical way that would help patients manage this multiplicity would be if hospitals were more readily able to share medical records and test results as Sandra explained:

Max’s notes are carried in various wheelbarrows. It will be great when it is available on one little stick. It would be really good to look it up on a system and see previous appointments. I think that will come in time. When things are more electronic, things will be much, much better. I think with his hearing and stuff like that, that would be really good because often the audiologists will ask me about his other tests and I won’t know the results. And things like Speech and Language would be able to see for things like swallowing. It would be really good! (Sandra, mother of Max, 11, CHARGE).

9.2.3 **Rarity**

The syndromes that we looked at are rare and participants knew that. Often patients knew more than GPs or local doctors and were pleased when they could speak to specialists who knew about their medical condition:

...we know that we are going to get, once a year, that people are going to understand. The full disability that we have got – they are going to understand. And not have oh what’s that. It’s just nice to be able to talk about it and for them to know what we are on about. (Jessica, 21, Wolfram).

However it was frustrating when patients attempted to tell professionals about their condition and were ignored. Patients and parents often know more than most doctors; they need to be listened to:

They never listen to the parents... They need to understand that parents know more about their children than some of the doctors do. We learn about it, we learn how to cope with it, we don't like being told to sit in the corner and shut up. We are intelligent people. We take the time to learn about the child's conditions. We learn how to communicate with the child. We learn everything that is needed to be known and we don't like being told that we're talking out of our rear ends! (Sue, guardian of Isla, 8, BBS).

9.2.4 Individuality

The experience of being a patient with sensory impairments and a rare syndrome and attending clinics is different for everyone. The manifestation of the syndromes is different in every case and people have individual needs relating to communication, mobility, and information, which all need to be met. These needs may also change over time.

Patients also have more things going on than their rare syndrome, both medically and in terms of their general life. Isla (8) as well as having BBS and epilepsy was also receiving bereavement counselling. Simeon (46, Usher) talking about having dyslexia, dyspraxia, and anxiety said, "We [have] got Ushers but there is so many things on the side that we have."

Patients too are much more than their syndromes or a sensory impaired person. For example Aalia (12, BBS) who for the majority of the interview had kept silent and did not contribute much regarding her thoughts and feelings on her experience of being a patient, was able to tell the researcher that at school she liked, "swimming, PE, cooking, art, computers." Angie, Paul, and Wendy also provide three further examples of the individuality of people with rare syndromes that cause sensory impairments:

I'm a Christian. I like swimming. Going to the gym is important for fitness. My friends. My family are very important to me – husband and daughter. One of the most important things for me is making things better for disabled people – campaigning, raising awareness.

That is extremely important to me. Making other people's lives I hope easier in the future. (Angie, 43, Stickler).

I like chess. I like 60s, 70s, and 80s pop music. I like psychedelic rock. I like sport listening to sport. I go swimming. I'm also doing hydrotherapy as well at the moment. I like going to the cinema and the theatre and going to the pub when I've got some money. I've been looking for a partner for the last 12 years! (Paul, 49, Alstrom).

Luke is an absolute thrill seeker if you had to put his number one on the list it would be to go on the biggest rollercoaster at Thorpe Park or Chessington and go on it for 3 hours! He loves swimming, trampolining anything that gives him the sensory feedback that he doesn't get. (Wendy, mother of Luke, 11, CHARGE).

We are glad to end this section by showing a more rounded picture of the individuals who talked to us.

10 Conclusion and Good Practice

10.1 Conclusion

The research project over more than a year, gathered information from a variety of sources, literature, patients, observations and audits, which, brought together, has produced a body of in-depth, detailed knowledge about what it is like to be a patient with a rare syndrome and sensory impairment. In some ways there is nothing new in the findings; people with visual impairment need help with finding their way, people with hearing impairment need consideration with communication, but this is here presented and brought together in terms of patients, not clinicians.

From this study data, has been drawn together to answer the research questions, to show in detail what patients feel and think as they attend hospital clinics, and from this to see what it is that distinguishes specialist clinics from others, and what features of the nationally commissioned clinics are distinct from those of other clinics. Using all of this data, it is possible to draw on factors which make the delivery of clinical services effective and efficient.

We started this project with three questions;

- How do people with sensory impairment (and their families/carers) experience their attendance at clinics in relation to their rare syndromes?
- What are the factors in relation to clinic type, environment, and attitude that affect this experience?
- What makes the delivery of services efficient and effective for people with sensory impairments and their families, across and between clinics, and what factors might inhibit this?

Sensory impairment causes difficulties with mobility, with communication, and with access to information. In attending clinics then, people with rare syndromes and sensory impairment do experience difficulties with getting to clinics, and getting around within them. In some places these difficulties are minimised by well defined

visual environments, by parking arrangements, and by staff who will guide patients when needed. They are also minimised by co-ordinated appointments so that patients do not have to attend too often. People with rare syndromes and sensory impairment experience difficulties with communication with staff at clinics. These include, for example, staff not using good communication practice for people with a hearing impairment, such as looking and speaking clearly, no provision for specialist communication modes, and a lack of time to understand what is happening and has been said. In some places these difficulties are minimised by people taking time, using hearing loops, speaking to the patient themselves, whether or not they are using an interpreter, and using clear speech. People with rare syndromes and sensory impairment experience difficulties in getting information before, during and after their clinic visits. Information was provided in formats they could not read, or was spoken in voices they could not hear, and even apparently simple activities, like knowing when they were called to be seen by a clinician, caused anxiety and tension. In some places these difficulties were minimised by the use of digital formats, staff who would read information out loud, or the use of text messages.

People with rare syndromes and sensory impairment often have many medical appointments, and are dealing with a variety of different symptoms and manifestations of their conditions. They experience difficulties with feeling isolated, because no-one else understands what it is like to be them, with having to make many medical appointments, taking time from school/work, and travelling long distances, with many, sometimes opposing, interventions and treatments. They can be confused and overwhelmed by multiple dealings with hospital professionals. Specialist, holistic clinics can help to minimise these difficulties by providing key liaison personnel, who have individual relationships with the patients, by co-ordinating care or at least providing a co-ordinated look at the care patients are receiving, and by ensuring that appointments and interventions are timetabled to be co-ordinated, this minimising both risk and travel/waiting time. Isolation can be reduced by arranging for patients (if they wish) to meet each other and share their experiences.

Thus through the study, we saw and discussed much good practice in relation to patients with rare syndromes and sensory impairments. Examples from patients indicated what was not working well and thus gave rise to more ideas about what good practice would be. What is most noticeable is that it is not necessarily major infrastructure changes or expensive organisational issues which would be most beneficial; some very minor changes in practice – for example in how patients are called for appointments – could make huge differences to patients' actual experience.

On the following pages we have outlined some Good Practice features, under three broad headings; Good practice in the environment, Good practice by staff, and Good practice in clinical situations.

They are divided into two sections, the first section includes items which might require authorisation, expenditure, systems reorganisation or other large scale thought and work. The second section however requires much less in terms of infrastructure, although it might involve training, and is more concerned with simple, and inexpensive changes which can be undertaken by people who are prepared to make an effort.

10.2 Good Practice Guidelines

Good practice in the environment

Policy strategies

- Buildings are easy to navigate, using colour coding of areas, clear and consistent signage, multiple formats of information
- There is good even lighting throughout corridors and in cafés and toilets
- There is at least clarity about car parking – such as patients only having to pay for what they have consumed (such as payment on exit)
- Department names are kept the same throughout the site (so not both “eye clinic” and “ophthalmology”)
- Toilets for people who need to lay down to be changed are made available
- All consulting rooms to have four walls and a solid door

Operational strategies

- Reception staff should offer directions and support for wayfinding on arrival
- Lights are kept on and additional lighting is provided in dark areas
- More play/activity equipment is provided in waiting areas, particularly thinking about the needs of older children, and also children who require specific sensory toys. TVs with subtitles and signing, and wifi are made available.
- Deliberate and clear paths are kept across waiting spaces – perhaps marked with tape/paint as clear paths
- Toilet leaflets are provided - simple directions for how toilets work, which are offered to patients (see Nina in 8.2.4)

Good practice in staffing

Policy strategies

- All staff, including reception and nursing staff, are trained in issues around sensory impairment, communication techniques and guiding
- There is a review and assessment policy for each hospital which includes a walk through and overview of procedures and clinics – such as 15 Steps, with a focus on sensory impairment issues
- Liaison personnel are provided; non-medical staff who can point patients in the right direction for both medical information, procedural information, and further support
- Training for staff in understanding the issues of dilation (vision reduction) for all patients, but particularly those who are deaf/ have a hearing impairment

Operational strategies

- Use of a sticker or colour-coded protocol or similar which outlines patients' communication and information needs and which all staff **read**. This is now a part of the Accessible Information Standard and will need to be implemented by July 2016
- All staff, reception and clinical, regularly check communication needs - 'Is that communication OK for you? Can I ask to you to repeat that so I know you understand?'
- Time is always given for genuine listening to patient – for those with rare syndromes only they can know the individuality of their conditions
- All staff always introduce themselves, clearly, including their job title, and this information is available in written (print, email, braille) format both before, and after appointments
- Staff give patients time, understanding that patients may have travelled a long way and have waited a long time
- Staff look at patients when talking, do not cover their mouths and pay full attention to patients during discussion
- Staff talk to patients, not interpreters or companions

- Staff ask if patients would like them to read information leaflets/forms/documents to them, and assist in filling out forms
- A culture of asking three key questions;
 - How can I help?
 - Am I getting this right?
 - What else can I do?

Good practice in clinic practice; appointments and procedures

Policy strategies

- Multiple formats of leaflets/information are always available (large print, audio, braille, electronic) and given to patients rather than leaving them to be noticed by patients
- Co-ordinated appointments are offered to minimise pressure on patients and families – either for families (syndromes often run in families) or multiple conditions (so that audiology and cardiology are carried out on the same day, or are on the same day for siblings)
- Procedures requiring anaesthesia are grouped to minimise the number of anaesthetics given
- Formal arrangements are made for patients to be able to meet by having ‘syndrome’ days for patient appointments
- Informal arrangements are encouraged for patients to meet each other – perhaps co-ordinated on social media, but a room is provided
- Loop systems are always available, working (checked) and used
- A quiet, private room is available for discussion with the receptionist, clinician or other

Operational strategies

- Appointment and post appointment information is available in accessible formats, for individual needs; always checking, is this format still Ok for you?

- Appointment and post appointment information is always available in digital formats if this is preferred
- Patients are given clear advance information about what the appointment is for; and what might happen as a result of it (patients were not always clear and some are juggling so many appointments, they have to prioritise)
- A plan of what exactly will happen at appointment or clinic is provided in advance, with who they will see, to minimise confusion
- Text messages are used to aid communication – e.g. to confirm appointments or tell people when to return from the café or outside if clinics are running late
- Information is promptly given in appropriate formats about waiting times; reasons for waiting; text messages are sent about delays
- There is a clear system for ensuring that people know when they are called to an appointment once in clinic, including approaching people directly if they need this
- Staff willingly talk to patients if they have difficulty with automated systems (e.g. booking in machines)
- Specialist clinics (e.g. ophthalmology, audiology) understand the needs of their own patients- e.g. vision impairment or hearing impairment
- There are smaller areas (with screens or similar) for children who find noise and bustle difficult
- Staff are ready to guide people to rooms when they are called and will ensure that someone will take them back
- Staff are always willing to provide, afterwards, written feedback on what was said in an appointment, in appropriate formats, especially digital information
- Named contact for further information following the clinic visit

11 Appendix

11.1 Wolfram syndrome

Wolfram syndrome was first described by Don Wolfram et al in 1938, who identified eight siblings in a family, of whom four had diabetes mellitus and optic atrophy. Three of the four patients went on to develop sensorineural hearing loss, and two developed bladder difficulties. Wolfram syndrome is also known as DIDMOAD (Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy, and Deafness) due to it being characterised by these conditions (Kumar, 2010). Juvenile diabetes mellitus and optic atrophy are the minimal criteria for establishing diagnosis (see Naderian et al, 2010). The prevalence of Wolfram syndrome is 1 in 770,000 in the UK (Barrett and Bunday, 1997).

Patients generally have diabetes mellitus followed by optic atrophy in the first decade (Barrett and Bunday, 1997). Diabetes mellitus is treated with insulin injections, blood testing, and by following a healthy balanced diet and getting regular physical activity.

Optic atrophy is when the optic nerve, or the cells within it, are damaged. The optic nerve sends images from the eye to the brain. It causes colour blindness and gradual loss of vision over time. Patients often begin by losing their peripheral vision (Kumar, 2010).

Cranial diabetes insipidus tends to appear in the second decade of life (Barrett and Bunday, 1997). Cranial diabetes insipidus is when the posterior pituitary gland, just below the brain, does not make enough of the hormone vasopressin which regulates the water in the body, so the body cannot concentrate urine. Symptoms include passing large quantities of urine often, and extreme thirst. It can be treated by replacing the hormone with a nasal spray or tablets. Catheterisation may also be necessary.

Sensorineural deafness happens because of damage to the hair cells (cilia) in the cochlea or auditory nerve. Individuals often have difficulty hearing high frequencies, and may be helped by the use of a hearing aid. Naderian et al (2010) suggest that hearing loss may be present in

48% of patients. Barrett et al (1995) suggest that high-frequency sensorineural deafness is usually diagnosed in the teens or 20s, and was seen in 62% of patients examined.

As well as the conditions mentioned above Naderian et al (2010: 53) state, “urinary tract atony, ataxia... mental retardation [sic] and psychiatric disorders are additional findings present in the majority of patients.” Barrett and Bundey (1997: 838) note that “Death occurs prematurely, often from respiratory failure associated with brainstem atrophy.” Most patients eventually develop all complications of this progressive, neurodegenerative disorder.”

Wolfram syndrome is an autosomal recessive condition which means that both parents need to have a copy of the faulty gene to pass it on to their children. If both parents have the faulty gene there is a 1 in 4 (25%) chance that their child will have Wolfram syndrome. There is a 2 in 4 (50%) chance that their child will be a carrier of Wolfram syndrome but not be affected by it, and a 1 in 4 (25%) chance that a child would be neither a carrier nor have Wolfram syndrome. Boy and girls are equally likely to have Wolfram syndrome.

11.2 Alström syndrome

Alström syndrome was first identified by Carl Henry Alström in Sweden in 1959 (Alström et al, 1959). Alström syndrome is a very rare condition with only 30 – 40 families in the UK affected, according to the NHS National Specialised Commissioning Team.

The whole of the body is affected in Alström syndrome with cone–rod retinal dystrophy, sensorineural hearing loss, obesity, insulin resistance, and type 2 diabetes mellitus. The onset and extent to which people are affected differs (Joy et al, 2007). Heart problems (dilated cardiomyopathy) are often seen in infants between 6 and 12 weeks of age, however unusually, if treated promptly and rigorously it is likely that the individual will recover from their cardiac problems (Paisey, 2014). People with Alström may also have high blood pressure, thyroid problems, bladder difficulties, liver and lung problems, fertility problems, short stature and problems with their bones and joints.

The first sign of Alström syndrome in individuals is nystagmus (uncontrolled movement of the eyes) and photophobia (sensitivity to light) in the first few months of life (Paisey, 2014). As the child develops visual loss is likely to occur as a result of cone-rod retinal dystrophy (Marshall et al., 2007). Cone-rod dystrophy is where first the cone cells in the eye, followed by the rod cells are lost. Cone cells help with central vision, and seeing colours and details, and rod cells help with peripheral vision and night vision.

Research suggests that hearing loss varies between people with Alström (Paisey, 2014). Progressive, high frequency, hearing loss is often noted in the first 10 years of life; however in a minority of cases hearing loss is not noted. If there is a hearing loss it may progress to a severe or moderately severe hearing loss by the age of 20, but for some people the hearing loss may remain stable. Hearing aids may be useful for some people, and cochlear implants may also be useful in a small number of cases (Paisey, *ibid*).

Like Wolfram, Alström syndrome is an autosomal recessive condition, which means that both parents must pass on a copy of the gene to their child, and males and females are equally likely to have Alström. The range of the phenotype also shows wide variations (Paisey, 2014). This means that Alström syndrome is likely to be different for different people e.g. some people may be able to read print for their whole life, whereas others will lose their sight completely.

People with Alström syndrome are not more likely than the rest of the population to have learning difficulties (Paisey, 2014), although “some reports indicate delayed psychomotor and intellectual development” (Marshall, et al., 2007: 1193).

Unfortunately the life span of patients with Alström Syndrome rarely exceeds 40 years, but early diagnosis and intervention can slow down the progression of the disease, and improve the length and quality of life for patients (Marshall et al., 2007).

11.3 Stickler Syndrome

Stickler syndrome was first described by Gunnar Stickler in the USA in 1965 (Stickler et al., 1965). He originally named it 'hereditary progressive arthro-ophthalmopathy' however it has since become known as Stickler syndrome.

Stickler syndrome is a group of inherited conditions which affect connective tissue (collagen). Collagen makes up a major part of connective tissue, and is a supportive tissue of the organs of the body. Sometimes connective tissue acts like a glue or binding, in other areas it acts like scaffolding. It also helps the muscles of the body stretch and tighten. Collagen is also found in cartilage which covers the ends of joints and is found in parts of the eye.

Depending upon which genes are involved, Stickler syndrome can be an autosomal dominant condition which means that only one copy of the mutated gene is needed, rather than two, to have the syndrome and that a person with Stickler has a 50% chance of passing on the syndrome to their child, however in a smaller number of cases it can be recessive meaning that a copy of the faulty gene is needed from both the mother and the father in order to have Stickler. Once again females and males are equally likely to have Stickler. Stickler is characterised by a distinctive facial appearance, eye problems, hearing loss, and joint problems (Baker et al., 2011). These signs and symptoms vary widely among people who have Stickler syndrome.

There are six types of Stickler syndrome. The different types of Stickler syndrome relate to the genes involved. People who have Stickler syndrome are likely to be very shortsighted (high myopia), have cataracts, retinal detachment and loss of cells in the retina. People may also have problems with eating and drinking due to having a cleft palate, they may have smaller jaws and flatter noses, hearing loss, early-onset osteoarthritis, and problems with bones and joints resulting in pain and affecting growth (mild spondyloepiphyseal dysplasia) (Baker et al., 2011). Stickler syndrome is the commonest cause of inherited and childhood retinal detachment (Fincham, et al., 2014).

In a study by Acke et al (2012), examining the data from 46 studies on people with Stickler syndrome, they found that a hearing loss was reported in 62.9% of people with Stickler syndrome, of this most (67.8%) had sensorineural hearing loss, 14.1% had conductive hearing loss, and 18.1% had mixed hearing loss. Mixed and conductive hearing loss was mainly found in younger patients and those who had cleft palate problems. However, it may also be undiagnosed.

The prevalence of Stickler syndrome has been estimated at between 1 in 7,500 to 1 in 9,000 (Baker et al., 2011).

11.4 CHARGE syndrome

CHARGE syndrome was first identified by Hall, and Hittner et al., both independently in 1979 (Deuce et al., 2012). Pagon et al. (1981) were first to use the term CHARGE, which was originally an acronym representing the diagnostic criteria (Coloboma, Heart malformation, choanal Atresia, Retardation of growth and/or development, Genital anomalies, and Ear anomalies). However, as understanding of the condition has developed, the current main features of diagnostic criteria refer to problems with the eye (mostly coloboma - where there is a gap in the structure of the eye), choanal atresia (when the nasal passages are blocked by bone or tissue), cranial nerve anomalies, and ear anomalies (Deuce et al., 2012).

CHARGE syndrome is a sporadic autosomal dominant condition, although there are a few incidences of parents who have mild forms of CHARGE, passing it on to their children. The incidence of CHARGE is between 1 in 10,000 and 1 in 15,000 births (Deuce et al., 2012). As with the syndromes already mentioned, the ways in which CHARGE affects people, and its severity, is different from person to person. As children with CHARGE get older, a number of features have been noted including curvature of the spine (scoliosis), migraine, epilepsy, cataracts, retinal detachment, delayed/arrested puberty, progressive hearing loss (Kirk, 2013).

Ear abnormalities not only affect hearing but also balance and, accompanied by low muscle tone, as well as a visual impairment, may mean that people with CHARGE have difficulty walking, or may need to

position themselves in 'unusual' positions, or perform repeated actions, in order to know where they are in their environment (Brown, 2005). Likewise, many people with CHARGE have problems associated with eating and drinking and this may be associated with having a cleft palate, paralysis in the face, as well as sensory issues (ibid).

People with CHARGE syndrome do frequently present with learning delay or disability, which may be related at least partly to the effects of dual sensory impairments. However, some people with CHARGE are University graduates.

People with CHARGE often have damage to multiple sensory systems, including smell and tactile senses as well as visual and hearing difficulties. They frequently have difficulty with balance and knowing and controlling their bodies in space. This, potentially coupled with learning and communication difficulties, means that a person with CHARGE may respond with "obsessive compulsive behaviours, rigidity, withdrawal, and frequent outbursts (Ranzon, 2001)" in Hartshorne (2013:2). Behavioural challenges can be made worse by sleep difficulties which affect many people with CHARGE (Heussler, 2013).

Behavioural challenges are common (but not always present) for people with CHARGE syndrome (Hartshorne and Cypher, 2004).

11.5 Usher syndrome

Usher syndrome is an autosomal recessive condition, with the predominant features being sensorineural deafness and progressive vision loss due to Retinitis Pigmentosa (RP). Usher syndrome was first described by Dr von Graefe in Berlin in 1858 with the name coming from Dr Charles Usher in the UK in 1914 (Millan et al 2011). The exact prevalence of Usher syndrome is unclear however Hope et al in 1997 suggested it might be as high as 6.2 per 100,000, and Sense suggest as high as 15 per 100,000. It may affect as many as 5% of those who are recognised as deaf in childhood (who are, probably, most likely to have Usher 1 or 2) (Kimberling et al., 2010; Dammeyer, 2012b). Women and men are equally likely to get Usher syndrome and people with Usher syndrome are no more likely than the general population to have learning difficulties.

Sensorineural deafness is due to damage to the hair cells (cilia) in the cochlea or the auditory nerve. Retinitis Pigmentosa is a gradually developing disease of the eye, which begins with loss of peripheral vision (to the rod cells) causing a limited visual field and nightblindness, but for people with Usher Syndrome, it can often (but not always) continue to an eventual deterioration of central vision (cone cells), usually in later life (Millan et al 2011). Côté, Dubé, St-Onge and Beauregard (2013: 140) talking about type 2 Usher, suggest that the visual impairment “may progress slowly or quickly, be stable, or degenerate until the person is completely blind.” As with the syndromes mentioned above, the severity and the ways in which people are affected by Usher syndrome depends from person to person.

Three ‘types’ of Usher are usually described (and within these subtypes exist) – Usher 1, Usher 2 and Usher 3 (e.g Bitner Glindzicz and Saihan 2014, Dammeyer 2012a). Typically the three types are used to differentiate the severity of deafness (Usher 1 connoting severe-profound deafness, Usher 2 moderate-severe deafness) the age of onset of vision difficulty (Usher 1 in early teenage years, Usher 2 in later teenage and Usher 3 in middle age). Usher 3 is then described as including progressive deafness (not so for Usher 1 or 2) and Usher 1 and sometimes Usher 3 includes balance difficulties (Millan et al., 2011).

11.6 Bardet-Biedl syndrome

Bardet-Biedl syndrome is named after George Bardet and Arthur Biedl who in the 1920s identified individuals who shared retinal degeneration, post-axial polydactyly (extra fingers and/or toes on the outside of the foot or hand) and obesity (Beales and Forsythe, 2014).

Extra fingers and/or toes, as well as fused or shortened digits, may be the only features of BBS which are present at birth. However within the first decade of life (around 6-8 years old) vision problems begin to occur, with a gradual loss of vision as time goes on (Beales and Forsythe, 2014). This is due to rod-cone dystrophy, where the rod cells in the eye, followed by the cone cells are lost.

Obesity is a very common feature of the syndrome and the development of type 2 diabetes mellitus is also prevalent. Developmental delay is

seen in a number of patients – but not all (Beales et al, 1999). Furthermore kidney problems and genital abnormalities are a feature of BBS and frequent monitoring is necessary. Conductive hearing loss is seen in around 20% of patients (Beales et al, 1999).

Bardet-Biedl syndrome is traditionally thought to be autosomal recessive although there may be some variations (Beales and Forsythe, 2014). In the UK the prevalence is estimated as 1 in 160,000 people (Forsythe and Beales, 2013).

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