Birmingham Centre for Rare Diseases

Realising the vision of high quality coordinated clinical care integrated with Research in partnership with patients
Birmingham Centre for Rare Diseases (CfRD)

One integrated Centre across 2 Sites

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Transition Lead
Consultant Nephrologist
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Rare Disease Lead for BCH
Renal Research and Transition Lead
Consultant Paediatric Nephrologist
Birmingham Children’s Hospital
Centre for Rare Diseases

Overview

• Background
• Developing a RD Clinical service of excellence
• The service, warts and all
• Optimising Research opportunities
• Future opportunities

What it is?

A (Supra) regional supportive structure delivering clinical excellence and research opportunities for patients with Rare Diseases
Strategy for Rare Diseases
Developed with patients

Expert Centres

- Coordinated care
- Adequate caseload
- Not dependent on single physician
- Transition
- Engage with patients
- Research

Accepted by NHS England +
DH Implementation Strategy 2018

- England (NHSE) & Home Countries

**Achievements and delivery**
- empowering those affected by rare diseases
- identifying and preventing rare diseases
- diagnosis and early intervention
- coordination of care
- the role of research

UK Rare Disease Policy Board - meeting the 51 commitments.
Rare diseases

- Rare diseases: important cause of illness and death
- Rare diseases affect many systems in the body
- 80% of rare diseases have a genetic component
- Often rare diseases are chronic and life-threatening
Why Centre for Rare Diseases in Birmingham?

- Supra-regional Centre serving 10% England, rich ethnic mix
- Cohesive clinical, multi-professional, cross organizational structure, Charity support, Managerial
- Developed & tested innovative RD Clinical care model with patients
- Strong innovative Clinical IT platform, HBRC-biobank
- Major existing research infrastructure-NIHR-Wellcome/UoB/BCH/BWH, TRC, GMC
- Leading young adult care
Common Diseases are now Becoming Rare

- Genomics and/or stratification by phenotype or response
- Stratified medicine
- Small (rare) sub cohorts of common disease
Extra-Renal Manifestations

Cystinosis

Reduced sweating & tears
Heat intolerance
Dental issues
The CfRD Clinical model

Governance remains unchanged

**CfRD Hub**
One stop Clinic
- Diagnostics
- Multi-speciality
- Multi-professional
- Transition
- Patient support

**Disease Registry**

**Spoke** (close to home)

**Intelligent IT Linkage**
- Hub & spoke interaction
- Remote Consult (MyHealth)
- Care pathway (Disease Specific)
- Rare Disease Website

**BHP NIHR**
(R&D Strategy)

**Birmingham Children’s RD**
• Transition

**Patient Support Groups**

**Pharma**
Centre for Rare Diseases in ITM at QEHB

Clinical engagement

- Cardiac
- IMD
- Liver
- Gastro
- Radiology
- Renal
- Clinical Immunology
- Endocrine
- Plastics
- ENT
- Clinical Chemistry
- Respiratory
- Clinical Genetics
- Rheumatology
- BCH Specialities
CfRD What’s in?

- Extensive Ophthalmology
- Dental Chair
- USS, Fibroscan, Echo
- Pharmacy: Research
- Bio banking sample collection
- Pharmacokinetics
- Resource Room
- Genomics: 100K
Phase I Clinical Services

- IMD including general metabolic, LSD, Alstrom
- LMBBS
- Renal genetic/renal metabolic/Cystinosis
- Renal transition
- Sjogrens
- IIH
- Liver, including liver/renal, liver/metabolic, Wilson’s and Liver Transition
- ENT, including NF2, paragangliomas and acoustic neuroma
- Ophthalmology (Birdshot chorioretinopathy, punctuate inner chorioretinopathy, ...)
- Neurology (Metabolic muscle disorders, neuro-genetic disorders, ion channel disorders)
- Cardiology (HCM)
Phase II & III
Elements of Clinics in CfRD

- Respiratory (Alpha I Anti-Trypsin)
- Liver II (Auto-immune hepatitis, PCB, PSC, NCPH, NAFLD, CF, Porphyria, Hereditary Amyloid, Neuro-endocrine tumours)
- Joint Rheumatology/Renal Lupus
- Cutaneous Lymphoma
- Neuro II
- Ophthalmology II
- Cardiology II (aortopathies, univentricular heart, Inherited cardiac transition, myopathy with cardiomyopathy)
- Plastics/Radiology –vascular anomalies
- Rare Immunology
- Tuberous Sclerosis
- Scleroderma
- Wolfram
CfRD: Activity

- Opened September 2015
- 20,000 attendances
- 71 separate clinics (>200 diseases)
- Strong patient/carer support/approval
- Bio banking
- Registries
Inherited Cardiac Conditions Patient Registry

Patient Summary

Consent

Receptive to participating in studies? Date Indicated Receptive/Not Receptive
No
28/09/2015

Consent Forms

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<th>Registry/Study</th>
<th>Consent Date</th>
<th>Withdrawn Date</th>
<th>Withdrawn Reason</th>
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<th>Withdraw</th>
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+ Add
Centre for Rare Diseases

The Centre for Rare Diseases is a brand new facility situated in the Heritage Building on the Queen Elizabeth Medical Campus in Edgbaston, Birmingham. Its vision is to develop a supra-regional integrated multi-specialty, multi-disciplinary centre of excellence for care of patients with rare diseases based within the Institute of Translational Medicine (ITM) (notably Translational Medicine). The focus is on highly organised one-stop clinics where patients (with their carers and visiting other pre-planned diagnostic tests and see all relevant specialists and the multi-disciplinary team at one visit at UHBT as the hub of their care.

Institute of Translational Medicine (ITM)

In 2012, Government funding was awarded for the development of an ITM Institute of Translational Medicine (ITM) in Birmingham in collaboration with the University of Birmingham. This takes a single point of entry for individuals, organisations and business seeking to maximise biomedical research translation for the benefit of the industry, economy and patients. It will facilitate the rapid assessment of cost-effective new drugs and medical devices within clinical settings. The ITM will be opened in June 2015. The ITM Clinical Research Facility will be operational from 1 September 2015.
Hello Sam

My Health Today
How are you feeling today?

Who can see my profile?

Edit profile
Contact the system administrator
Visit my bio

You are signed up to receive
You are signed up to receive

Recent Activity
Page: 1 of 1
Last refresh: 12/12/2013 11:27

There is no recent activity to report.

My profile
Edit your profile and personalise your settings.

Support network
Test Pass-support
@ myhealth@QEH
Support

Manage
Expand support

My records
Communicate
Help
My favourites

Medication
View, update and add to your hospital medication.

Results
View, analyse and update your results.

External blood results
Submit your blood results to the hospital.

Inpatient history
View and record your hospital visits.

Calendar
View your hospital appointments and create appointments and reminders of your own.

Contacts
Manage your hospital contacts.

My vault
Upload files and share them with the hospital.

Support network
2 in your support network

Gadgets
Blood pressure results
We have no recorded results for today.
Submit today's results below.
Systolic:
Evaluation and Research

• Key performance indicators regularly monitored by project group

• Patient and clinicians evaluations have been performed

• The system is part of a number of research projects
Research Opportunities:
‘Go-to place for RD Research’

• 1st dedicated comprehensive 1 stop clinical adult RD centre
• Develop existing & new large RD Cohorts
  – Integrated Registry development
  – Integrated bio-banking & GMC
• Build on extensive RD research activity
• Birmingham: NIHR RD Paediatric cross cutting
• Leverage IT platform: Myhealth (Virtual research networks) & PICS
• ERNs
• Patient Groups support & QEHB Charity Support
RD Centre Facilitates Research

- NIHR Bio-repository site
- 2 clinical services
- Leverage pharma and investigator-led research
- Support of UoB Industry engagement team

<table>
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<th>Speciality</th>
<th>Grant Values</th>
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<td>Rheumatology</td>
<td>226K</td>
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<tr>
<td>Neurology</td>
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<td>IMD</td>
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<td>Dermatology</td>
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<td>Gastroenterology</td>
<td>28K</td>
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<tr>
<td><strong>Total</strong></td>
<td><strong>2.5 million</strong></td>
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Pharma and CfRD?

- Benefits to Pharma
  - One stop portal for industry to access
  - Existing large RD Cohorts based on clinical excellence
  - Embedded research structure
  - Strong IT platform on which to build/Follow up
  - Trial acceleration model may well make drug development viable

- Everyone wins!
Research & Educational Opportunities

CfRD ‘Go-to place for RD Research’

- Pharma interest, one stop access
- Potential to accelerate drug assessment
- Access to large patient cohorts
- RD is the key focus of Birmingham Health Partners
- Reduce costs by economies of scale
- Rare Disease trials acceleration potential
Future Developments

- Remote consultation
- Trials acceleration programme
- Enhanced registry development (NLP)
- Financial and governance structure to underpin
- Enlargement
- Enhanced bio-banking
- Greater Pharma linkage
- Basic science
myVirtualClinic

myhealth@QEHB is piloting dedicated virtual clinics for some of our patients with long term conditions, and also those patients who live far away. myVirtualClinic provides patients with direct face-to-face contact with their named healthcare professional for specific consultation appointments. Advice and treatment planning is arranged with the patient in the same way as an outpatient appointment.

Please provide your contact no. here since your healthcare professional may need to call you if a technical fault occurs during the virtual appointment. 01213717323

Upcoming appointments

24 May 2017 - Dr J Ferguson Virtual Clinic, HEPATOLOGY, Liver Medicine

Appointment time: 10:30 AM

You can join the virtual appointment from 10:00 AM, once joined please wait for your healthcare professional to become available.

Expected appointment duration: 15 minutes

Prepare for my appointment  Add to personal calendar
Audio recordings are made available for Clinicians and Patients
Rapid assessment of novel therapies: a global challenge in Rare Diseases

• The last decade has seen a revolution in diagnostics and therapeutics which is transforming patient outcome

• As a consequence rapid and accurate assessment of novel therapies is now an urgent medical priority

• Assessment of novel therapies is slow and this global deficit represents a major challenge to pharma

• Particular challenge in Rare Diseases
Key TAP Milestones

- Initial TAP award: Nov 2010 (36 months)
- Official TAP launch: 30-Sep-2011
- First TAP trials launched
- First TAP trial to fully recruit
- 14 TAP trials opened in 4 years
- Initial funding period
  - 12-month funding extension awarded
- 5-year programme funded
Our Overall Vision: Integrating rare diseases delivery and translational research

Life Science Model for Birmingham (BHP)

- Children's Services
- Adult Services
- Women's Services
- University

Stratified and Personalised Medicine

Integrated R&D and Service Delivery
Thank You

STAR APPEAL
Lighting the way for children with rare diseases

Queen Elizabeth Hospital Birmingham Charity