Developing the 1st Children’s Rare Disease Centre

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By your side
Rare Diseases in the West Midlands

- Population 6 million people
- Young population 1/5 under 16 yrs of age
- Multicultural with large ethnic mix
- Stable population
- We treat more than 9000 patients with over 500 different rare diseases per year
Birmingham Women’s & Children’s NHS FT

- Most subspecialties (34) on single site
- 270,000 patient visits/year
- Largest Paediatric Intensive Care in Europe
- Nationally Designated Specialist Centre for several services
- WM Regional Genetics Services largest in UK
- Outstanding CQC rating!
1st Children’s Rare Disease Centre @ BCH

- One stop access to MDT clinics
- Bespoke MDT clinic space
- Family rooms, support and peer networking space
- Supported by Patient Organisations
- Access to Registries and Research Projects
- Video conferencing for virtual follow up reviews
- Fully integrated transition: prenatal to adults
- Co-designed with children, young people & families affected by rare diseases
Family days (rare & undiagnosed conditions)
Collaborative approach
100K Genome Project
- Rare Disease Registry
- NIHR Common & Rare Disease Biobank
- NIHR CYP-Tech: Medical Technology Innovations for Children & Young People affected by Rare Diseases
- Rare Disease Service Delivery
- Recruitment for clinical trials
- Transition
The 100K Project: BCH leading recruiter for Rare Diseases & Cancer

Recruited over 3000 participants

Start of Personalised Medicine & Future Research Opportunities
• Natural history of Rare Diseases often not known
• Essential for clinical trials
• Difficult to identify patients with RD by using hospital data systems
• Important information for Hospital, NHS Commissioners, and Research
• Linking up with PHE NCARDS and other hospitals
CYPmedTech Initiative

- Children’s and Young People medical technology group
- Founded from the NIHR’s MIC call to build expertise and capacity in the NHS and to develop new medical child health technologies
- Leading paediatric NHS organisations act as centres of expertise, bringing together patients, clinicians, researchers, commissioners and industry
- CYPmedTech is the only dedicated children's technology network

CYPmedTech is a collaborative partnership dedicated to the development of technology specifically for the healthcare of Children and Young People in paediatric specialty areas where disease burden and mortality risk is high and in highly specialist fields that may otherwise not receive extensive technology focus
The Rare Disease Theme priority areas:

- Accelerated diagnosis
- Information and support for patients
- Improved coordination of care
- Transition from child to adult
- Technology to support knowledge
- Identify unmet needs
- Pull together diverse stakeholders
- Support and develop new medical technologies
- Work with founders to fund diverse and unique calls within rare disease umbrella

What’s Next?

In Leeds on June the 6th we will be holding an event with the Translate group (www.translate-medtech.ac.uk) and the TITCH network, this will focus on identifying problems within rare disease and working on solutions to these problems with academics and industry.

Website coming soon, in the mean time follow us on Twitter @cypmedtech or contact us at nihr-cyp-mic@nihr.ac.uk and keep an eye on www.titch.org.uk for updates.
Where would technology make a real difference within rare diseases?

- By improving communication between the patient and care team
- By improving accurate diagnosis – quicker, easier and more streamlined
- By supporting the seamless transition to adult services
- Be more science focused enabling new med tech to aid with treatment
- Be more app and software focused to help with care management

Do you have an unmet need in rare diseases you would like to talk to us about? Contact us at nihr-cyp-mic@nihr.ac.uk
Rare Diseases: exciting future

- New knowledge & technology
- 1st Children’s Rare Disease Centre at BCH
- West Midlands unique setting for rare disease research
- Rising to challenges in rare diseases through research
- Collaboration vital

Thank you