Paediatric Rare Disease Clinic

Comisiwn Bevan Commission

Dr Torsten Hildebrandt, Consultant Paediatrician
Mrs Sian Townsend, Paediatric Matron, CTMUHB

Contact: torsten.hildebrandt@wales.nhs.uk

Project Background:

Rare disease, defined by an incidence of less than 1 in 2000 people are individually rare, but collectively common. Currently there are up to 7000 different conditions recognized, a number which will increase in the future. 1 of every 17 of us will be affected by a rare disease at some time in our life. Children are disproportionately affected with 75% of rare diseases presenting in young age. Rare diseases often are life limiting and /or life threatening. Patients and families often have complex care needs with significant impact on education, independent lifestyle, financial and social security, physical and mental health. All of this can lead to healthcare inequality and a lack of provisions including the delivery of timely, tailored emergency management, appropriate surveillance and long-term support.

Project Aims:

- Develop a new multidisciplinary service for children with rare disease in partnership with patients, parents and carers
- Identify children with complex rare disease in the catchment area of a Paediatric unit in a District General Hospital
- See these patients in clinic and address care needs as above

Project Approach:

The Paediatric Department at the Princess of Wales Hospital Bridgend serves as pilot for the proposed service.

All paediatric patients under current care were reviewed. Patients fulfilling the key criteria of having a disease with an incidence of less than 1:2000 were highlighted (161 patients).

The Congenital Anomaly Register and Information Service (CARIS) kindly provided data of all registered paediatric patients up to May 2023 (935). Patients with systemic or multiorgan disease were identified (102 patients).

Paediatric Consultants notified patients to the project regularly.

Data as above was cross referenced identifying 125 eligible children with 103 different conditions.

Project Outcomes:

The service for children with complex rare disease has been established and is running with following objectives:

- Multidisciplinary clinic run in partnership with patients and their carers
- Enabling patients and their carers to 'understand their disease'
- 2-hour appointment offered
- Pre-clinic extensive disease-based search of databases and medical literature documented on Welsh Clinical Portal and discussed in clinic
- Surveillance needs and time intervals defined, documented and actioned
- Emergency measures defined and documented
- Information patients and carers bring to the appointment i.e. via social media discussed
- Patients signposted to patients' groups
- Referral and research centres identified and approached
- Transition to adult services initiated if appropriate
- Follow up communication via appointment, telephone or home visit as per patients' choice offered
- Service open for referrals
- 125 patients with 103 conditions identified and awaiting review
- 8 patients seen

8 patients seen within the service (February 2024)
Diagnosis:
1p36 deletion syndrome
16p11.2 microduplication syndrome
17q12 deletion / HNF 1B deletion RCAD
HNF 1B deletion
Neurofibromatosis type 1
Phelan McDermid Syndrome
PPP2R5D missense variant
Partial trisomy 11q, monosomy 15q26, unbalanced translocation

Project Impact:

Patient Reported Outcome Measures (PROMs) assess the quality of care delivered from the patient perspective. Patient reported experience measures (PREMs) are tools that capture a patient's experience and perception of what happened during their encounter. PROMs and PREMs were validated through CEDAR (Centre for Healthcare Evaluation Device Assessment and Research).

The clinic was started before PROMs and PREMs were agreed within the Welsh NHS structure and therefore completed for only 3 patients, showing an improvement in all categories. Analysis of outcomes is ongoing.

Next Steps:

- Increase patient numbers
- Collect meaningful feedback data from patients, parents and carers
- Collect meaningful feedback data from referrers to the service
- Expand service across 3 paediatric sites in CTMUHB

Key Conclusions:

The number of patients seen in the Rare Disease Clinic so far is small. Evaluation is ongoing with early results being promising with good feedback from parents and service users throughout.

Overall, it is hoped that there will be a shift in mindset dealing with rare disease. With 1 in 17 of us being affected, rare disease is common. No healthcare professional can have an overview over all rare conditions, but the tools to acquire knowledge at the time of use, interlink professionals, patients and carers are more accessible than ever. Dealing with rare disease will be part of normal paediatric practice to the benefit of our patients.