Cross Party Group for Rare, Genetic & Undiagnosed Conditions

Inaugural Meeting

Date: 02/10/2019, 12:00 – 13:30

Attendees:		**Apologies:**
Angela Burns AM (Chair)	Dr Alison May (Cardiff Friends of Sickle Cell & Thalassaemia)	Angie Contestabile (British Liver Trust)
Emma Hughes, Genetic Alliance UK (Secretariat)	Dr Annie Procter (Cardiff & Vale UHB)	Del Braim (Respiratory Innovation Wales)
Alan Thomas (Ataxia & Me)	Dr Amy Hunter (Genetic Alliance UK)	Naja Frenning (Patient)
Dr Jayne Spink (Genetic Alliance UK)	Dr Graham Shortland (Chair, Rare Disease Implementation Group & Cardiff & Vale UHB)	Michaela John (Genomics Partnership Wales)
Dr Samuel Chawner (Cardiff University)	Dr Karen Reed (Wales Gene Park)	Angela Burgess (Wales Gene Park)
Ellie Russell (Tuberous Sclerosis Association)	Faith Walker (Cardiff Friends of Sickle Cell & Thalassaemia)	Helen-Mary Jones AM
Joanne Ferris (ABPI Wales)	Lucia Elghali (Parent of child affected by Fragile X Syndrome)	
Marie James (Parent of adult affected by tuberous sclerosis complex)	Martin Williams (Parent of young adult affected by rare condition)	
Menai Owen-Jones (Pituitary Foundation)	Mike Bryan (Researcher, Angela Burns office)	
Prof Angus Clarke (All Wales Medical Genomics Service)	Prof Julian Sampson (Medical Genetics, Cardiff & Vale UHB)	
Prof Meena Upadhyaya OBE (Ethnic Minority Women in Welsh Healthcare)	Sondra Butterworth (Rare disease researcher and patient affected by thalassaemia)	

1) **Welcome & Introductions**

The groups Chair, Angela Burns AM welcomed stakeholders to the meeting and emphasised the importance of getting a group together focusing on the issues facing families affected by rare, genetic and undiagnosed conditions. Stakeholders gave brief introductions.

2) **Presentations**

Dr Amy Hunter, Genetic Alliance UK
Martin Williams, parent of a son who has recently been diagnosed with a rare condition following 13 years of have an undiagnosed genetic condition
Dr Samuel Chawner, Cardiff University.

<u>Dr Amy Hunter</u>

- Dr Amy Hunter highlighted findings from a report published by Genetic Alliance UK in 2018 focussing on over 2000 responses from rare disease patients and carers sharing their experiences of mental health problems. Having a rare condition had a huge impact on emotional wellbeing and mental health. 36% of patients and 19% of carers reported that they had thoughts about suicide. The impact of trying to get a diagnosis and poor care coordination also had a negative impact on respondents mental health. Poor care delivery was exacerbated by poor awareness of the condition by health professionals and not being believed or taken seriously by healthcare professionals.

- Evidence based recommendations that were delivered fell under two themes; empowering rare disease healthcare professionals and integrated mental health care with rare disease services. The recommendations were:

1. Healthcare professionals should be provided with the skills, knowledge and capacity to:
   -demonstrate awareness of the emotional challenges of living with a rare disease
   -handle discussions about mental health sensitively

2. Parents and carers should be routinely signposted to sources of support by professionals

3. Coordinated rare disease services should include assessment of mental health needs and access to mental health services. This should be extended to carers.

<u>Martin Williams</u>

- Martin spoke passionately about how living with his son's unknown genetic diagnosis for over 13 years had affected his family and specifically about his experiences of how it affected his mental health and the mental health of his wife.

<u>Dr Samuel Chowder</u>

- Dr Samuel Chawner spoke about some genetic conditions which lead to a range of overlapping needs in children. The IMAGINE ID research project assessed a broad range of mental health and clinical domains and took an approach which harnessed the expertise of

parents and children through parent reports, interviews and questionnaires and direct child assessments and teacher reports.

- The study included 258 children with one of thirteen relatively commonly diagnosed genetic conditions and 106 sibling controls. They found that there was an increased risk of psychiatric conditions with 79.8% meeting the criteria for a psychiatric condition.

- Another question looked at how clinical outcome changes with age. Traits which were more sever in older children included mood symptoms, psychotic experiences and peer relations which indicates that if early difficulties in childhood are left unaddressed they become more severe with age and highlights the needs for early support and intervention.

- Sam concluded that children with genetic conditions are one of the most vulnerable groups in society for developing mental health problems. They need better access to support and early intervention with a step change in clinical services being required. Common service pathways for genetic conditions as a group would be an effective step forward as would the creation of dedicated care pathways for rare childhood genetic conditions as a group**.**

3) Discussion

Following the presentations, discussion focused on the following themes:

a. consider how psychiatric support could be integrated into services as well as ensuring more support and training for staff

b. consider how transition services could be more flexible in respect of defining the age of transition and supporting the individual holistically

c. ensure support is based on individual need rather than diagnosis or age

d. include commitments to improve mental health planning and service provision in whatever follows the UK Strategy for Rare Diseases and Welsh Implementation Plan post 2020

e. i) Ensure that Rare diseases are a priority area in the National Clinical Plan for Wales and that full engagement takes place to develop high level clinical pathways for Rare Disease patients.

ii) Ensure that any changes with the current system of delivery/implementation groups, as the new Welsh NHS Executive body is set up, includes full consideration of Rare Diseases and appropriate resource /funding to transform and deliver services for Rare Disease patients in line with other groups.

4) AGM

Angela Burns AM was nominated as Chair of the grouo. The nomination was seconded by Janet Finch-Saunders AM. Sondra Butterworth and Dr Alison May nominated and confirmed Emma Hughes, Genetic Alliance UK as Secretariat.

5) Financial Information

No expenditure made from the meeting.

6) Date of next meeting

Provisionally agreed: 10 December 2019.