Cross Party Group for Rare, Genetic & Undiagnosed Conditions

Second meeting

Date: 14.01.2020, 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 Iolo Doull (Welsh Health Specialised Services Committee)	Angela Burgess (Wales Gene Park)
Nick Meade (Genetic Alliance UK)	Dr Karen Reed (Wales Gene Park)	
Anna Evans (Cystic Fibrosis Trust)	Dr Graham Shortland (Chair, Rare Disease Implementation Group & Cardiff & Vale UHB)	
Kate Learoyd (National Society for Phenylketonuria)	Lucia Elghali (Parent of child affected by Fragile X Syndrome)	
Rebecka Bow (Parent of child affected by cystic fibrosis)	Claire Rowlands (Member of NSPKU)	
Rick Greville (ABPI Wales)	Mike Bryan (Researcher, Angela Burns office)	
Chris Cook (Member of NSPKU)	Emma Steers (Member of NSPKU)	
Lowri O'Donovan (Cardiff University)	Dr Samuel Chawner (Cardiff University)	
Menai Owen-Jones (Pituitary Foundation)	Prof Meena Upadhyaya OBE (Ethnic Minority Women in Welsh Healthcare)	
Caroline Graham (Member of NSPKU)	Dawn Bowden AM	

1) **Welcome & Introductions**

The groups Chair, Angela Burns AM welcomed stakeholders to the second meeting of the CPG and introduced the speakers, firstly Kate Leoroyd from the National Society for Phenylketonuria, then Anna Evans representing the Cystic Fibrosis Trust and

Rebecka Bow, parent of a child affected by cystic fibrosis and finally Nick Meade from Genetic Alliance UK.

2) **Presentations**

Kate Leoroyd, National Society for Phenylketonuria
Ann Evans, Cystic Fibrosis Trust and Rebecka Bow, CF campaigner
Nick Meade, Genetic Alliance UK

Kate Leoroyd

- Phenylketonuria (PKU) is a genetic metabolic disease affecting about 1 in 10,000 births. The body does not metabolise an amino acid within protein.
- PKU needs careful management to prevent permanent harm to the brain.
- For the last 50 years the condition has been "treated" by not eating protein. This can be stressful, miserable and doesn't always work.
- There are new treatments available for PKU including Sapropterin (Kuvan) licensed in 2008. This medicine works in about 1/3 of PKU patients, depending on their genotype.
- Allows people to metabolise protein naturally, prevents brain damage
- Pegvaliase was licensed in 2019 and is currently used in Germany and the USA.
- The NSPKU have made submissions to the SMC in Scotland which failed in 2018 and a submission to NICE on Pegvaliase which has been paused. Submission to NICE for Kuvan has been stopped due to ongoing litigation. Also 3 submissions to make the medicine available through NHS England have failed.
- There was a detailed discussion with members from the NSPKU who are affected by the condition about how the condition, specifically the restrictive amount of food they are allowed to consume affects their lives.
- There was a further discussion about the impact of having a child with the condition and the burden on parents who are taught to do blood tests so they can check levels are correct. Officially it takes 19 hours/ week to prepare the food for a PKU patient. Time and the financial burden is not considered or valued. From a social perspective, the condition can mean that children feel socially excluded.
- As a result of newborn screening for PKU, there are approximately 3-4 new cases per year.
- Kuvan costs between £25,000 - £50,000/year per patient.

Anna Evans and Rebecka Bow

- Anna Evans spoke about the CF communities campaign to access Orkambi in the UK.
- In July 2016, NICE issued its final recommendations which was a decision for Orkambi not to be made available to patients in the UK. in the same month the CF Trust propose interim access agreement using the UK CF Registry,
- Between October to November, both England and Wales announced that patients would have access also.

- In June 2017, the community protested across all 4 nations. In October 2018, Wales extended access to Kalydeco. In January 2018, a petition was handed in at the Senedd.

- There was a Health and Social Care Committee enquiry in November 2018 and in September 2019, Scotland were the first country to allow access.

- Only 50% of the CF community have access to drugs due to many not being suitable for all types of CF. These are the first drugs to deal with the underlying cause of the condition and to stop the drowning effect of mucus in a patients lungs.

- The campaign belongs to the campaigners and Rebecka Bow stated that parents must pull together and fight for access.

- Some key learnings from the process:

  1. Very little support for patient communities following a negative appraisal
  2. Despite the infrastructure to support interim access there was no willingness to achieve this
  3. The support and skills campaigners bring are invaluable
  4. It can be difficult to navigate the lines between the NICE system and devolved power

Nick Meade

- Nick drew on examples that had been highlighted to show there is a systematic problem with access to orphan medicines in the UK. There are many other examples.

- 'Action for Access' is a report developed in response to a number of Genetic Alliance UK member organisations raising concerns over processes for accessing medicines in their communities. The UK systems for approving NHS funding for rare disease medicines means that fewer medicines gets to patients and decisions are slower.

- Due to NICE deciding to now appraise all new medicines from April 2020, the position for Wales will be different as there will be a mandate from NICE for drugs to be made available. There could also be fewer decision makers. There is a question about the capacity/ timing of appraising all new medicines and NICE are currently reviewing their methodology but with no timescale available for when this review will publish.

- Nick drew on the report published by Genetic Alliance UK, 'Action for Access' and recommendations about how the systematic problems of fragmentation, lack of flexibility, problem with capturing value, transparency and delays in the access to medicine landscape needs to be addressed. He drew on a third pillar approach when presenting the solution:

1. Evidence - patients should be able to use new medicines in the NHS to gather appropriate levels of evidence for appraisals

2. Systematic - a single, flexible rare disease NICE pathway

3. Money - Transparent discussion of key elements of current system

3) Actions

Following the presentations, action points arising from the discussion focused on:

1. Angela to meet with NSPKU and families

2. Parliamentary question regarding Kuvan and proposing a short debate on PKU in the Chamber

3. Follow up meeting between Angela Burns AM and Genetic Alliance UK to discuss the 'Action for Access' report

4**) Date of next meeting**

Provisionally agreed: 5 May 2020

Angela will also host a reception for Rare Disease Day 2020 at the Senedd on 25 February, 6-8pm