ALL PARTY PARLIAMENTARY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Undiagnosed Children's Day - the systemic barriers to diagnosis

14.00-15.00, Wednesday 27 April 2022
Portcullis House, Room R

Parliamentarians in attendance: Liz Twist MP (Chair)
Baroness Pauline Neville-Jones
Christina Rees MP
David Duguid MP

Speakers: Nick Meade, Joint Chief Executive & Director of Policy, Genetic Alliance UK
Marie Pritchard, parent representative from SWAN UK

Attendees:
Farhana Ali, Genetic Alliance UK
Rachel Clayton, Genetic Alliance UK
Rachel Rimmer, Rare Autoinflammatory Conditions Community - UK
Rhia Arden, MPS Society (The Society for Mucopolysaccharide Diseases)
Sarah Wynn, Unique
Simon Bull, Charcot-Marie-Tooth UK
Stine Holm, Office of Liz Twist MP
Matthew Rose, Muscular Dystrophy UK
Michaela Regan, Muscular Dystrophy UK

MINUTES

Liz Twist MP, Chair, APPG on Rare, Genetic and Undiagnosed Conditions

Liz Twist MP welcomed attendees to the meeting. Liz noted that Undiagnosed Children's Day will be taking place on Friday 29 April - to raise awareness for all who live with a syndrome without a name.

Diagnosis is one of the most important cross cutting issues for the rare condition community as diagnosis opens the door to services, treatment, care and support.

Many face significant challenges in getting a diagnosis, with more than a third of people with a rare condition having to wait more than five years. Approximately 6,000 children are born in the UK every year with a genetic condition likely to remain undiagnosed.

Marie Pritchard, Parent Representative at Genetic Alliance UK

Marie shared her experience of the diagnostic odyssey with her ten year old daughter Dottie who has an undiagnosed condition.

Having a child with an undiagnosed condition is difficult. For families, it can be very difficult to get healthcare professionals to listen to and respect your concerns. Sometimes, parents are even blamed for their child's developmental delays. The feeling of regret and guilt is often present in parents with a child with an undiagnosed condition as they don't know if they could have stopped it or may have even caused it.

Marie explained the emotional toll of having an undiagnosed condition. It has a significant impact on the mental health of the whole family. It can leave them feeling isolated from family, friends and healthcare professionals. It leaves questions about the future which is a source of worry and concern.

Hospital appointments became a large part of Dottie's life and the responsibility of coordinating care is also a burden on the family which can be overwhelming when Dottie is treated by a multidisciplinary team.

SWAN UK provided a support network for Marie and her family. It gives a sense of belonging and comfort to know that you are not alone. The community gives support and empowers families to continue during the journey to diagnosis.

Marie's experience with the 100k Genomes Project was mixed. The project provided some hope for a diagnosis and provided access to care that had previously been denied. However, communication between the healthcare professional and the patient needs improving. The possible outcomes weren't made clear, delivery of diagnosis wasn't well thought out, it wasn't made clear that just because a diagnosis hadn't been found yet your child could still have an undiagnosed genetic condition.

Marie emphasised the importance of receiving a diagnosis of a genetic condition. It provides access to services and support. It also provides some relief and can answer questions for the family.

Nick Meade, Joint Interim Chief Executive at Genetic Alliance UK

Nick introduced 'Good Diagnosis: Improving the experiences of diagnosis for people living with rare conditions'. The report identifies the eight guiding principles of a good diagnosis experience as being;

- accurate and timely
- informed and supported
- collaborative and coordinated
- respected and acknowledged

Central to a good diagnosis are healthcare professionals who recognise and are aware of rare conditions. We must urgently take steps to raise awareness of rare conditions among healthcare

professionals and to provide them with the information and resources required to support their patients.

People living with rare conditions describe feeling frustrated at poor communication about their care and the 'battle' and 'struggle' to get referred for tests, or for assessment at specialist centres. Even once a diagnosis is made, it may not be the end of the struggle. When an individual has a diagnosis of a rare condition, they may continue to experience delays in being started on the correct care or treatment pathway, they may experience delays in accessing appropriate services, medicines or research.

Having to fight for the right care or treatment can be extremely challenging, particularly when unwell. The fight can be harder when a person is unsure about what rights they have, or who can support them. It is vital that people with rare conditions are aware of what to expect from the diagnosis journey, aware of their rights and how they can challenge decisions.

The Good Diagnosis report recommends that a Rare Conditions Good Diagnosis Patient Rights Charter should be developed to clearly communicate the standard of care people with rare conditions should expect to receive.

Genetic Alliance UK will work to develop a Good Diagnosis Patient Rights Charter.

Discussion

Diagnosis doesn't always lead to a prognosis. Support groups like SWAN UK continue to provide much needed support even after diagnosis. The Good Diagnosis report is useful in bringing together what we already know: a diagnosis is a huge relief for families.

There are many things that can be learnt from the 100K Genomes Project in providing a good diagnosis but also how to provide support and information once they have a diagnosis.

Baroness Neville Jones detailed the value of care coordination. Care coordinators that know the case history and have expertise in managing these conditions, can help with appointments and therefore reduce the stress that families experience.

The NHS is beginning to understand the importance of catering to those who do not fit under common needs.

Guest speaker Marie Prtichard spoke about the importance of communication between hospitals when receiving care across multiple hospitals; there is value in having access to all health professionals in one place.

Michaela Regan noted the value of specialist centres and specialist nurses for care coordination was raised.

Liz Twist MP raised the point that for any progression in care to happen, patients and patient organisations often have to do a huge amount of work. Diagnosis isn't always a gateway for getting care and treatment.

Both Cristina Rees MP and David Duguid MP asked about Welsh and Scottish Action plans respectively and the opportunities for each nation to learn from the other actions plans.

It was noted that patient organisations support people to ask the right questions to access genome sequencing. Sarah Wynn commented that access is varied depending on your geographical location. Educating healthcare professionals is important but supporting families is also essential.