CROSS PARTY GROUP ON RARE, GENETIC AND UNDIAGNOSED CONDITIONS

Systemic Barriers to Diagnosis

12.00-13.30, Wednesday 18 May 2022
Online

Parliamentarians in attendance: Mike Hedges MS (Chair)
Russell George MS
Rhun Ap Iorwerth MS
Mark Isherwood MS

Guest Speakers: Claire Swan, Representative from SWAN UK
Dr Graham Shortland, Outgoing Chair of the Rare Disease Implementation Group
Natalie Frankish, Genetic Alliance UK's Policy and Engagement Manager for Scotland and lead on the Good Diagnosis project

Minutes

Nick Meade, Joint Interim Chief Executive and Director of Policy, Genetic Alliance UK, opened the meeting as Mike Hedges was delayed at another meeting.

AGM for the APPG on Rare, Genetic and Undiagnosed Conditions

The AGM for the CPG on Rare, Genetic and Undiagnosed Conditions was conducted. Mike Hedges was elected as Chair of the APPG. The following were elected as officers of the CPG:

- Russell George MS
- Rhun Ap Iorwerth MS
- Mark Isherwood MS

Rachel Clayton, Policy and Public Affairs Officer at Genetic Alliance UK was elected as the secretariat.

Systemic Barriers to Diagnosis

Claire Swan shared her experience of care for her child, Lucy, who has an undiagnosed condition. Claire noticed something different in Lucy when she began to miss development milestones.

When expressing her concerns to doctors, she found it difficult to be taken seriously and was often shrugged off as an anxious parent. Claire fought to advocate for Lucy and, once her symptoms were recognised, coordination of care was left to Claire and her partner. Both of Lucy's parents have had to leave work to become her full time carers.

"We suddenly had appointments coming at us from all directions and our family was thrown into a brand-new world of unknowns."

Claire expressed that the lack of prognosis meant uncertainty for the future for her family. Without a diagnosis, her family cannot understand how the condition might progress and what Lucy may need in the future.

This new world felt very lonely but Claire found friends who understood the challenges they were facing through SWAN UK, a support group for families with children with undiagnosed conditions.

Natalie Frankish introduced the report 'Good Diagnosis: Improving the experiences of diagnosis for people living with rare conditions' which highlights the experiences of the diagnosis journey for people living with rare conditions across the UK and considers how the experience of diagnosis can be improved. 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

The report also identified that awareness of rare conditions amongst health professionals is central to the experience of diagnosis, and that more must be done to ensure health professionals working in the NHS are equipped to support people with rare conditions.

The report put forward four recommendations:

1. The UK Rare Disease Framework Delivery Partners should consider developing a central repository (such as an online portal) of information on rare conditions for healthcare professionals.
2. People with rare conditions should be given a diagnosis care plan when they begin their journey to diagnosis.
3. People living with rare conditions in the UK should be offered access to a care coordinator throughout their journey to diagnosis.
4. A Rare Conditions Good Diagnosis Patient Rights Charter should be developed and included in each national action plan. The Charter should clearly communicate the standard of care people with rare conditions should expect to receive. The Charter should be based on the identified Principles of Good Diagnosis as defined by this report.

Genetic Alliance UK will be taking forward recommendation four and will be engaging with a wide range of stakeholders to develop a Good Diagnosis Patient Rights Charter in collaboration with the rare community throughout summer 2022.

Attendees were invited to participate in this project and those wanting to take part can contact natalie@geneticalliance.org.uk

Wales Rare Disease Action Plan

Graham Shortland updated attendees on the development of the Wales Rare Disease Action Plan, covering actions which address the four priority areas and underpinning themes of the UK Rare Diseases Framework and the recommendations set out in the CPG 2021 final report which states that:

- The Welsh Action Plan must include commitments to improve mental health planning and service provision .
- Transition services must be more flexible when defining the age of transition and supporting individuals.
- Access to orphan and ultra-orphan medicines.
- Impact of Covid-19.

The action plan is currently being finalised and is expected to be launched in June.

Discussion

Mark Isherwood MS raised the issue of third sector engagement post diagnosis. Increasing awareness amongst professionals of how to access charities for support and information once a diagnosis has been delivered is essential. He also asked how the voice of people

with rare conditions and support organisations are being included in the design, delivery and effectiveness of the Wales action plan.

Graham Shortland explained that the Wales Rare Disease Action Plan has been developed jointly with people living with rare conditions and support organisations representing them via multiple engagement events and workshops. Genetic Alliance UK, Wales Gene Park and The Genomics Project also hold a position on the Rare Disease Implementation Group (RDIG) responsible for developing the action plan and will continue to inform the work that will be undertaken to implement the plan.

Graham Shortland noted that the action plan is an ongoing document and there will be opportunities to change and update the plan should notable gaps be identified. It is fair to say that the work is not done and RDIG will continue to work closely with the rare condition community.

Cross border collaboration was discussed in terms of the SWAN Clinic and action five in the England Rare Diseases Action Plan to develop an undiagnosed clinic. Graham Shortland noted that Wales SWAN clinic was much more developed than the project in England but they would be happy to liaise with them to find potential areas of collaboration.

Attendees shared comments on the action plan, which were largely positive. There was support for the commitments relating to better coordination of care. Attendees considered the Wales Rare Diseases Action Plan to hold many opportunities for progress and noted that there will be challenges to its implementation, particularly as the recovery from the Covid-19 pandemic will have a significant impact on the health landscape, both in terms of pressure on resources and opportunities to change the way care is delivered.

The importance of research into rare conditions was also noted, and there was a request to improve patient involvement in the design of research and introducing a source of funding to enable this. .