Welsh Cross Party Group on Muscular Dystrophy and Neuromuscular Conditions

Meeting minutes

Wednesday 29th November, 10-11am

Virtual, via Zoom

Introduction

On 29th November 2023, the Welsh cross party group (CPG) for Muscular Dystrophy and Neuromuscular Conditions held an Annual General Meeting (AGM). The meeting also heard about the latest updates on newborn screening in the UK as well as the challenges facing neuromuscular services in Wales.

This document provides a summary of the meeting's discussions and actions. A list of meeting attendees is provided at the end.

About the CPG

This cross party group's purpose is to raise awareness of the need to develop specialist services for patients with muscular dystrophy and related neuromuscular conditions in Wales.

It plays an important role to raise awareness of muscle wasting and weakening conditions. It also promotes links between Parliament, individuals and families affected by the conditions, charities, health professionals, scientists and decision-makers.

Annual General Meeting

An AGM was held at the start of the meeting to elect office holders.

The following decided to stand down from their role(s):

- Rhun ap Iorwerth MS (Plaid Cymru, Ynys Môn) stepping down from the role of chair. The MS will continue as a member only.

The following were elected to new role(s):

- Huw Irranca-Davies MS (Welsh Labour and Co-operative Party, Ogmore) was elected to the role of chair.

The following were re-elected to their role(s):

- Muscular Dystrophy UK to continue as secretary for the CPG.

Summary of meeting

Agenda item 1: Welcome and introductions

1.1. Meeting chair and CPG chair Rhun ap Iorwerth MS (RI) welcomed participants. The chair introduced the meeting agenda and proceeded to the AGM.

1.2. No apologies for absence were noted.

Agenda item 2: Annual General Meeting

2.1. RI thanked attendees for the opportunity to chair the CPG and passed over responsibilities to Huw Irranca-Davies MS (HID). HID thanked RI for leading the group and reiterated the importance of there being a firm chair to take the CPG forward.

2.2. Responsibilities for chairing the meeting also then switched from RI to HID.

Agenda item 3: Update on newborn screening for rare diseases

3.1. On behalf of Muscular Dystrophy (MDUK), Rob Burley (RB) expressed MDUK were glad to be staying on board as secretary and congratulated the previous and incoming chairs.

3.2. RB noted MDUK's role as the co-secretary of the UK Spinal Muscular Atrophy (SMA) Newborn Screening Alliance, working alongside Alice Fabre (AF) and SMA UK.

3.3. RB introduced the UK All-party parliamentary group (APPG) on Muscular Dystrophy's inquiry at the end of 2022 looking at newborn screening for rare conditions. There are few conditions screened for in the UK compared to in Europe and in the US. The inquiry looked at the evidence required to have more conditions added onto the list of conditions screened for in the UK, with a particular focus on SMA. The inquiry was supported by the APPG on rare and undiagnosed conditions.

3.4. RB described the inquiry report's recommendations, published in May 2023.

- The approach taken by the UK National Screening Committee (UK NSC) to assessing conditions for newborn screening needs to be expedited. Whilst it should be robust, there are ways in which it could be more pragmatic.
- The criteria and evidence requirements for a condition to be accepted for newborn screening need to be reviewed so that they are fit for purpose for rare diseases.
- A clear and transparent approach focused on stakeholder engagement is key.

3.5. SMA was noted by RB to be a prime candidate for screening because of the availability of effective treatments by NICE. With treatments seen as a key motivator for screening, the landscape for other neuromuscular conditions is more promising than before, as more gene therapies are potentially coming on, including for Duchenne Muscular Dystrophy (DMD).

3.6. RB provided an update on the recently announced research study by Genomics England. This will cover over 200 conditions, including SMA and a few other neuromuscular conditions. The aim is to analyse the genomes of 100,000 newborn babies. RB noted this was not a pilot nor a screening programme, but that the findings of the study may help shape future decisions around screening. MDUK had heard that under the study, the turnaround time for results would be approximately 4-6 weeks, in contrast to the 7-8 days taken for the heel prick test used in screening programmes. This is why it is so key that conditions be included in the UK newborn screening programme.

3.7. RB noted that currently, genomic screening is not at a stage to be rolled out more widely in the UK. The technology is still very much at a research stage.

3.8. AF introduced the UK SMA Newborn Screening Alliance, which is a group of patient organisations, clinicians and academics, all focused on securing screening for SMA at the earliest opportunity.

3.9. AF described the significant progress made in prognosis for patients. Before the arrival of new treatments, SMA was one of the leading causes of child death. It is therefore

important that these treatments are administered before symptom onset, because otherwise the damage is irreversible. The fastest route to this is via newborn screening.

3.10. AF discussed the growing case for screening. For example, there are now videos showing the stark difference treatment can make to the lives of individuals, in contrasting siblings who did and did not get access to these new treatments. Screening for SMA has already been modelled and is shown to be highly cost effective.

3.11. AF updated attendees on the latest plans for SMA screening in the UK. The UK NSC want to produce an economic model and conduct an in-service evaluation of how screening works across the NHS in the UK. The UK NSC are now scoping the evaluation, with the first meeting of the oversight group taking place (including all four nations and relevant ministerial teams). The next meeting will open out to broader stakeholders. This group will make decisions about what the evaluation needs to do, how expansive the programme is, and what is needed for it to get up and running.

3.12. In response to audience questions, a discussion took place about the changes in screening for DMD in Wales over the years. Limited quality assessment capacity was a key reason why it was dropped. Since screening was abolished, DMD is being picked up much later. There have been various discussions to advocate for newborn screening for DMD because of the emerging treatment landscape. RB noted the UK NSC reviewed DMD last year. Although better management is available, the lack of immediately available treatments was a sticking point. MDUK had flagged that the UK NSC needed to rethink their screening decision and are in touch with them to work on this.

Agenda item 4: Resilience and sustainability of neuromuscular services in Wales

4.1. David Heyburn (DH) provided a state of play of the condition of local neuromuscular services. It is well known that neuromuscular services have been in a fragile state over many years. DH referred to this as an ‘own goal’ for government and the NHS. There have been multiple attempts to try and secure resources to improve services, but to date these have been unsuccessful. In one of the latest such attempts: over the past 12 months, service specifications were pulled together, workforce needs were assessed, and a hub and spoke model thought out (centralised teams in three regions with specialised know how and skills to treat complex cases, as well as support for general services with moderate to mild disease cases).

4.2. In principle, health boards recognise the situation is not optimal, but they note that many other services are in a similar situation. DH recognised the reality that neuromuscular services are a small red blip on radars that have pretty big red fires.

4.3. DH also noted that neuromuscular services cross over many specialities and that finding the data to demonstrate harm/potential harm is therefore very difficult. This makes it more challenging to pitch what is needed when the context is that multiple services are experiencing pressure.

4.4. Business cases have gone to executive teams and health boards, and follow up conversations have been asked for. DH noted that the bill of around a million pounds was not an insignificant sum to consider. Next steps include looking at phasing and prioritisation, as well as considering the possibility of including neuromuscular services in boards’ Integrated Medium Term Plans, so as to not get lost off the radar.

Agenda item 5: Open discussion and questions

5.1. Attendees noted the backdrop to this discussion, where no area, including beyond health, that is not under pressure. So even getting small amounts is quite a challenge. It is also challenging to keep this on the agenda, from a politician's point of view, when there are so many fires to put out.

5.2. Audience members with lived experience raised the changes in screening for DMD in Wales and the need to get back to earlier diagnosis. RB was asked about the global research study for early screening for DMD by Parent Project Muscular Dystrophy (PPMD) and how much MDUK was working with PPMD. RB agreed with the need to re-engage with PPMD and learn from international experience, which the UK NSC was noted to be a little against.

ACTION: MDUK to follow up on learnings from international experience for DMD screening.

5.3. Health professionals in the audience noted it was essential that services in northwest Wales be improved and asked what was being done to prioritise this. In the southwest and southeast, there had been investments from a local point of view. But in north Wales, there was a lack of infrastructure and support for the single person working there on their own. DH shared those concerns. There have been conversations with different chief executives on trying to see what support can be provided. But DH acknowledged a team was needed to really manage this and health boards were struggling to understand what this means. It was sometimes a struggle to be heard and there was no easy answer. DH was continuing to speak with colleagues and trying to find their way into the right conversations to press on. Attendees raised MDUK's lobbying role in these discussions. RB agreed in principle, but in light of previous experience noted the importance of finding the best moments to advocate.

ACTION: MDUK to pick up conversations with DH and colleagues about plans on how to improve regional services.

5.4. HID initiated a discussion on where the CPG can add value. Experience from other CPGs is that these groups tend to be helpful in adding a nudge and pressure.

5.5. The role of neuromuscular networks was considered. The neuromuscular network needs to be seen as an operational delivery network, which would give it a mandate and anchored position in the wider structure. This would not improve the workforce situation overnight, but would ensure neuromuscular services do not drop off people's radar. There were also questions around where the neuromuscular network sits in regard to a neurological conditions network. The CPG could add weight for the central all Wales wide NHS executive to have these conversations.

5.6. RB agreed for part of the role of the CPG to be about bringing people in to hold them to account, as well as to be sharing information. DH's experience of the CPG reflects some of the wider challenges facing the system. For example, there had been previous attempts to hold chief executives to account, but these fell short when they did not turn up to meetings. DH agreed to work together to plan more of these to try again.

ACTION: MDUK to work with CPG members and others to identify the best ways to use the CPG, including to influence at executive and health board levels.

5.7. RB referenced the meeting of the Scottish cross-party group on Muscular Dystrophy last week. There were discussions around mapping service provision in Scotland and comparing that with the landscape for similar conditions, for example Motor Neurone Disease and Parkinson's disease. This would be a way of drawing attention to local issues and conversations around how to improve equity could be picked up in the Scottish CPG.

ACTION: MDUK to consider producing a policy report around the provision of neuromuscular services in Wales.

5.8. Attendees discussed how conditions could be added more quickly onto the list of conditions screened for, for example through shortening the time between treatments being approved and screening programs being implemented. RB noted horizon scanning was one of the recommendations in the UK APPG's inquiry report. Lots of Integrated Care Boards and providers were just not ready for treatments being approved by NICE because everyone was working on day-to-day needs and there was not enough budget now for longer term requirements. The capacity implications of new treatments were not small either. For example, around five years ago, there was one treatment approval process for one neuromuscular condition that MDUK was involved in. There are now thirteen across five conditions.

Agenda item 6: Summary and closing remarks

6.1. RB noted MDUK's ongoing work to understand the issues people with lived experience of muscle wasting and weakening conditions wanted MDUK to focus on. A community survey is now open until late December. The results may also inform the CPG's programme of work.

ACTION: MDUK to share MDUK's community survey with meeting participants.

6.2. HID acknowledged some of the challenges in delivering change for neuromuscular services in Wales and reiterated the value of the CPG in bringing people together to identify pressure points.

6.3. HID thanked presenters, attendees and the previous group chair. The meeting was then closed.

List of attendees

Members of the CPG:

- Rhun ap Iorwerth MS (Now former chair of the CPG).
- Huw Irranca-Davies MS (Now current chair of the CPG).
- Rob Burley (Director of Campaigns, Care and Support at Muscular Dystrophy UK; CPG secretary).
- Lucia Gillespie (Advocacy & Information Officer for Wales, at Muscular Dystrophy UK; CPG secretary).
- Farhan Khan (Health Policy Manager at Muscular Dystrophy UK; CPG secretary).

Non-CPG members/invited observers/guest speakers included:

- Rhys Hughes (office of Rhun ap Iorwerth MS).
- Alice Fabre (Project Manager at UK SMA Newborn Screening Alliance).
- David Heyburn (Head of Operations at Public Health Wales; Deputy Chair at Wales Neuromuscular Network).
- Heledd Tomos (Physiotherapist at NHS Wales).
- Joanne Oliver (Neurological Conditions Strategic Clinical Network manager at NHS Wales Executive).
- Carolyn Middleton (person with lived experience).
- Gareth Middleton (person with lived experience).
- Sylvia Evans (person with lived experience).
- Kenneth Lewis (person with lived experience).
- Janet Neilson (person with lived experience).
- Lucy Patten (person with lived experience).
- Richard Haselgrove (person with lived experience).
- Richard Enos (person with lived experience).
- Paul Magness (person with lived experience).
- William Silcox (person with lived experience).
- Neil Thorneycroft (person with lived experience).
- Welsh language interpreter from the Senedd.