



All-Party Parliamentary Group for Muscular Dystrophy: AGM and Meeting on Newborn Screening for Rare Conditions

Summary of Meeting

Wednesday 24th May 2023, 4pm – 5pm

Room O, Portcullis House, and 'Hybrid' via Zoom

Background

On Wednesday 24th May 2023, the All-Party Parliamentary Group (APPG) for Muscular Dystrophy held an Annual General Meeting (AGM) and launched their latest report on Newborn Screening for Rare Conditions – see Appendix for a summary.

This document provides an overview of the meeting's discussion, a summary of key actions, and an attendance report.

AGM of the APPG for Muscular Dystrophy

The APPG for Muscular Dystrophy plays an important role to raise awareness of muscle-wasting conditions. The APPG promotes links between parliament, individuals and families affected by the conditions, charities, health professionals, scientists, and decision-makers.

The APPG for Muscular Dystrophy held their AGM during this meeting. Attending Officers of this APPG voted for a Chair and for Vice Chairs. The result of this AGM vote confirmed the following roles:

- Mary Glindon MP (Chair, North Tyneside, Labour).
- Liz Twist MP (Vice Chair, Blaydon, Labour).
- Steve Brine MP (Vice Chair, Winchester, Conservative).
- Jim Shannon MP (Vice Chair, Strangford, DUP).
- Baroness Thomas of Winchester (Vice Chair, Life Peer, Liberal Democrat).

Overview of Meeting

This meeting began with welcomes and introductions, and then included an AGM and a structured panel-style discussion of the report as well as contributions from attendees.

Overview of the Report: Newborn Screening for Rare Conditions

- Rob Burley (Director of Care, Campaigns, and Support, Muscular Dystrophy UK) provided an overview of the APPG for Muscular Dystrophy's Inquiry and Report on newborn screening for rare conditions. He also thanked all those who contributed to the inquiry and the report.
- On the background to this inquiry and report, he emphasised that newborn screening is essential for receiving a faster diagnosis.
- Rob stated that challenges remain to have rare conditions added to the newborn screening list. He referenced the fact that only nine rare conditions are screened for in the UK Newborn Screening Programme (UK NBC) compared to over 20 in Europe, and over 50 in the USA.
- After outlining the aims of the inquiry and report – noted in the Appendix – Rob Burley defined the report's recommendations:
 - The approach taken by UK NSC to assessing conditions for newborn screening needs to be expediated. 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.
- He argued that there is more to do and called for the UK Screening Committee to engage with the APPG for Muscular Dystrophy and Muscular Dystrophy UK (MDUK).

The patient impact of newborn screening

- Portia Thorman (Advocacy Lead, Spinal Muscular Atrophy UK) spoke of her personal experience as a parent of a child with Spinal Muscular Atrophy (SMA). She stated that SMA is not screened for in the UK.
- From her personal experience, she noted that while the first symptoms of SMA presented in her child at 5-weeks old, they received a diagnosis at 4-months old. She argued that this diagnosis was too late, noting the extent of care and equipment her child requires.
- Portia highlighted the economic impact of this late diagnosis to both the NHS and education system. She stated that the nursing care for her child costs the NHS approximately £460,000/year which does not include any of the equipment, feed, and physio. She also highlighted that she gave up her career as a teacher to become a carer.
- She called for SMA to be added to the UK Newborn Screening Programme.

The role of patient groups on newborn screening

- Rachel Clayton (Senior Policy and Public Affairs Officer, Genetic Alliance UK) presented on Genetic Alliance UK's work on newborn screening and welcomed this report.
- Rachel provided an overview of Genetic Alliance UK's work to bring together and advocate on behalf of over 200 organisations that support people living with rare and genetic conditions in the UK.
- She noted the importance of newborn screening for rare conditions, including the need for a timely and accurate diagnosis. She cited evidence that more than a third of people with a rare condition wait more than five years to receive a diagnosis.

- Rachel also stated that newborn screening for rare conditions is essential to ensure the provision of timely treatment, appropriate care, and support to minimise the impact of a condition.
- She argued that a key priority that the rare disease community's voice is heard to inform national level decision-making.

Open Discussion: Effectiveness of the UK Newborn Screening Programme

- Attendees expressed frustration and inadequacy at the fact that only nine rare conditions are currently screened for in the the UK Newborn Screening Programme.
 - APPG Members reflected these frustrations, stating the slow progress in screening for rare conditions in the UK.
 - One attendee argued that the UK Newborn Screening Programme is a 'laughingstock' not a 'gold standard' compared to socio-economically similar countries.
- Attendees discussed the evidence requirements for rare diseases, emphasising the need to understand the nuances and expertise of rare diseases.
- Attendees argued for reform of the UK Newborn Screening Programme to ensure more rare conditions are screened for.

Open Discussion: Cost and Evidence

- Attendees noted the importance of expanding the newborn screening programme and the cost-benefit.
- One attendee argued that the cost of treatment and care for a patient with SMA, for example, could help to fund the screening for all current newborns for SMA.
- It was argued that expanding rare conditions screened for in the newborn screening programme would not only ensure faster diagnosis and benefit patients and their families, but also benefit the NHS and wider economy.

Open Discussion: Awareness

- Attendees called for greater public awareness and an open discussion on the challenges with the current UK newborn screening programme.
- Attendees asked what more could be done to secure greater awareness from the public to increase pressure on the Government to reform the newborn screening programme.
 - One attendee argued that its key to galvanise Doctors, GPs, and other Healthcare professionals on newborn screening and to build alliances.
 - Attendees also noted the importance of patient groups working collaboratively to secure support in the press.
- Members of the APPG supported the tabling of a Westminster Hall Debate in Parliament on newborn screening for rare conditions and expressed support for corraling wider support from MPs.
- Members of the APPG emphasised the importance of engaging with the Secretary of State for Health and Social Care, and the Minister for Social Care with responsibility for rare diseases. Members expressed their willingness to write to these Ministers on behalf of their constituents.

- Members of the APPG also referenced the current Health and Social Care Committee inquiry on Prevention, noting that there is scope for attendees to submit responses on newborn screening and diagnosis for rare diseases.

Actions

The following Actions were agreed at this meeting:

- **Members of the APPG for Muscular Dystrophy** to table a Westminster Hall Debate on Newborn screening for rare diseases – **June 2023**.
- **Members of the APPG for Muscular Dystrophy** to write to the Secretary of State for Health and Social Care on the UK Newborn Screening Programme and rare diseases – **ongoing**.

Attendance Report

Attendees of this meeting included:

- Mary Glindon MP (Chair of the APPG for Muscular Dystrophy, MP for North Tyneside, Labour).
- Steve Brine MP (Vice Chair of APPG for Muscular Dystrophy, MP for Winchester, Conservative).
- Liz Twist MP (Vice Chair of APPG for Muscular Dystrophy, MP for Blaydon, Labour).
- Jim Shannon MP (Vice Chair of APPG for Muscular Dystrophy, MP for Strangford, DUP).
- Rachel Clayton (Senior Policy and Public Affairs Officer, Genetic Alliance UK).
- Portia Thorman (Advocacy Lead, Spinal Muscular Atrophy UK).
- Liz Ryburn (Support Services Manager, Spinal Muscular Atrophy UK).
- Dr Lucy McKay (Chief Executive, Medics 4 Rare Diseases).
- Georgina Morton (Founder and Chairperson, ArchAngel MLD Trust).
- Pat Roberts (Programme Director for Newborn Screening, Arch Angel MLD Trust).
- Jonathan Gibson (Policy and Public Affairs Officer, Metabolic Support UK).
- Alice Fabre (Project Manager, UK Spinal Muscular Atrophy Newborn Screening Alliance).
- Michaela Regan (Public Affairs Partner, Roche).
- Professor Bobby Gaspar (CEO, Orchard Therapeutics).
- Dr Mark Atherton (Specialist Registrar in Paediatric Neurology, Sheffield Children's NHS Foundation Trust).
- Nick Mills (Advanced Nurse Practitioner, Sheffield Children's NHS Foundation Trust).
- Sian Ball (Occupational Therapist, Sheffield Children's NHS Foundation Trust).
- Professor Tracey Willis (Consultant Paediatric Neurologist, NHS RJA).
- Dr Corinna Clark (Research Fellow, University of Warwick).
- Dr Catherine McWilliam (Consultant Clinical Geneticist, Ninewells Hospital).
- Trevor McLeese (Patient representative).
- Rob Burley (Director of Care, Campaigns, and Support, Muscular Dystrophy UK).
- Citta Widagdo (Health Policy and Commissioning Officer, Muscular Dystrophy UK).
- Amanda Haest (Senior Account Executive, WA Communications).

Appendix – Overview of the Report: *Newborn Screening for Rare Conditions*

Newborn screening is a fundamental tool to help achieve timely diagnosis of rare conditions. Over recent years, the advancement of access to treatments and other health support has increased the importance of timely diagnosis for people living with neuromuscular conditions and other rare conditions.

In October 2022, the APPG on Muscular Dystrophy launched an inquiry into newborn screening for rare conditions and the evidence requirements for the acceptance of a condition onto the national newborn screening programme. The inquiry was supported by the APPG on Rare, Genetic and Undiagnosed Conditions.

The APPG inquiry aimed to understand:

- The views of people living with rare conditions on the potential impact of newborn screening on families, society and the NHS.
- The types of evidence that should be considered to allow robust but timely decision-making about adding rare conditions to the UK national screening programme, and how uncertainty arising from evidence relating to rare conditions should be handled by the UK National Screening Committee (UK NSC).

The inquiry showed support for the UK NSC to continue to take a robust approach to assessing the appropriateness of conditions for newborn screening. However, there was consensus that there was scope for a significantly faster, more transparent approach that takes factors such as the progressive nature of many rare conditions, and the significant associated health impacts for babies born with them, into greater account.

The inquiry report defines recommendations into three overarching themes:

- The approach taken by 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.

The APPG for Muscular Dystrophy report “Newborn Screening for Rare Conditions” is available online [here](#).