Inquiry into the UK National Screening Committee (NSC) and the NHS newborn blood spot screening programme

Submitted by ArchAngel MLD Trust, MPS Society, and MLD Support Association UK.

We propose an inquiry into whether the NSC and the process for adding new conditions to the newborn screening programme are fit for purpose and improve public health by preventing avoidable death and suffering.

The programme has fallen behind.

- One condition added in ten years (ten conditions total), well behind Norway (39), Poland (36), the Netherlands (31), Denmark (25), Germany (23) and France (16)[i], which averaged ten additions over a similar period.
- The NSC process does not work, including consistently requiring In-service-evaluations (or similar).
- Has failed to keep up with modern treatments, risking public confidence.

The inquiry should span rare diseases where early identification is essential, though the problem is exemplified by the NSC's recent recommendation against MLD screening.

- Considering the same evidence, criteria and international standards, the UK is the outlier: Norway[ii], the USA[iii], Canada[iv] and Sweden[v] have recommended screening.
- The stated reasons (that screening does more harm than good, treatment won't improve outcomes, and cut-offs aren't established) are heavily disputed by experts and other jurisdictions' outcomes.
- The NSC reviewed just 3 of 25 pieces of evidence it deemed credible[vi].
- The treatment is effective, holding NICE approval[vii].
- Norway's screened over 80,000 newborns without issue[viii]. Multiple pilots confirm screening works[ix].

What evidence should the Committee collect?

- Why the UK lags comparable countries (one positive recommendation in ten years, despite the same criteria, evidence and standards).
- Time taken to review and recommend conditions: the NSC has considered SMA for eight years[x] (20+ European countries began screening in this time[xi]), SCID for thirteen[xii].
- The human impact of screening fewer conditions than peers: 2-3 children born with MLD, SMA, SCID, MPS I and ALD a week - all screened elsewhere[xiii] but not in the UK[xiv].
- The disconnect between treatment and screening approvals.
- The NSC's accountability, governance, transparency and performance, including whether its processes suit rare diseases and how underperformance is identified and addressed.

What we aim to achieve

A programme that keeps pace with clinical progress and international peers, so fewer children miss timely diagnosis of rare, treatable conditions. The inquiry should test whether the NSC's outcomes and processes are fit-for-a-modern screening system and recommend reforms where they fall short or where the government's early-diagnosis intentions go unmet.

Suggestions for witnesses

- Rare-disease patient-organisation representative - Georgina Morton, Chair, ArchAngel MLD Trust.
- Representative of the Newborn Screening Collaborative - Giles Lomax, CEO, SMA UK.
- Clinical expert (rare disease) - Dr Arunabha Ghosh, Clinical Research Fellow in Paediatric Inherited Metabolic Diseases, Central Manchester University Hospitals NHS Foundation Trust.
- Laboratory expert (newborn screening) - Teresa Wu, Director of the Willink Laboratory.
- International screening expert - Andreas Øberg, Senior Consultant at Oslo University Hospital.
- NSC representative.
- DHSC representative.

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[i] Time to decide: Learning from international approaches to newborn screening decision-making', Genetic Alliance (2025) https://geneticalliance.org.uk/wp-content/uploads/2025/07/Time-to-decide-Learning-from-international-approaches-newborn-screening-decision-making_Highres-3.pdf.
[ii] Øberg et al. 'Nationwide Newborn Screening for MLD in Norway: First-Year Implementation and Performance', poster presented at WORLDSymposium2026, San Diego, February 2026.
[iii] US Department of Health and Human Services, 'Secretary Kennedy Adds Duchenne Muscular Dystrophy, Metachromatic Leukodystrophy to Newborn Screenings', 16 December 2025. Available at: https://www.hhs.gov/press-room/secretary-kennedy-adds-duchenne-muscular-dystrophy-metachromatic-leukodystrophy-to-newborn-screenings.html (accessed 15 June 2026).
[iv] Canada's Drug Agency (CDA-AMC) 2026. Health Technology Review - Newborn Screening for Metachromatic Leukodystrophy. https://cda-amc.ca/sites/default/files/hta-he/HC0079-NBS_MLD_Combined_Review.pdf.
[v] Socialtyrelsen 2025. Nationella riktlinjer 2025: Screening för metakromatisk leukodystrofi (MLD) Rekommendation om att utöka nyföddhetsscreeningen - Remissversion. https://www.socialstyrelsen.se/publikationer/nyfoddhetsscreening-for-metakromatisk-leukodystrofi-mld-2025-10-9823/.
[vi] UK National Screening Committee, Newborn screening for metachromatic leukodystrophy (MLD): a rapid evidence review (Kleijnen Systematic Reviews Ltd, November 2025, published January 2026), pp. 85-87. https://view-health-screening-recommendations.service.gov.uk/document/66f8195b-f544-4e27-ae0f-d665a257679e/download.
[vii] NHS England, 'NHS to roll out life-saving gene therapy for rare disease affecting babies', 4 February 2022. Available at: https://www.england.nhs.uk/2022/02/nhs-to-roll-out-life-saving-gene-therapy-for-rare-disease-affecting-babies/ (accessed 15 June 2026).
[viii] Øberg et al. 'Nationwide Newborn Screening for MLD in Norway: First-Year Implementation and Performance', poster presented at WORLDSymposium2026, San Diego, February 2026.
[ix] 1. Hong X et al. Genet Med. 2021;23(3):555–561; 2. Kelly NR et al. Mol Genet Metab Rep. 2023 Dec 14;38:101037; 3. Wu THY et al. Mol Genet Metab. 2024;142(1):108349; 4. Laugwitz et al N Engl J Med. 2024 Oct 3;391(13):1256-125; 5. Malvagia S et al. Int J Neonatal Screen. 2025 April 24;11(2):30.
[x] UK National Screening Committee, *Spinal muscular atrophy* recommendation page, https://view-health-screening-recommendations.service.gov.uk/sma/ (accessed 15 June 2026).
[xi] Pane M et al., 'Spinal muscular atrophy in the era of newborn screening: how the classification could change', *Frontiers in Neurology*, (2025). Available at: https://www.frontiersin.org/journals/neurology/articles/10.3389/fneur.2025.1542396/full (accessed 15 June 2026).
[xii] Time to decide: Learning from international approaches to newborn screening decision-making', Genetic Alliance (2025) https://geneticalliance.org.uk/wp-content/uploads/2025/07/Time-to-decide-Learning-from-international-approaches-newborn-screening-decision-making_Highres-3.pdf.

[xiii] Time to decide: Learning from international approaches to newborn screening decision-making', Genetic Alliance (2025) https://geneticalliance.org.uk/wp-content/uploads/2025/07/Time-to-decide-Learning-from-international-approaches-newborn-screening-decision-making_Highres-3.pdf.
[xiv] UK National Screening Committee. *UK NSC recommendations*. Available at: https://view-health-screening-recommendations.service.gov.uk/ (accessed 15 June 2026).