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Professor Sir Mike Richards
UK National Screening Committee
39 Victoria Street
London
SW1H 0EU
uknsc@dhsc.gov.uk

Dear Professor Sir Mike Richards,

Newborn screening for Metachromatic Leukodystrophy (MLD): Summary and Recommendations document following the publication of the decision of the UK NSC to reject newborn screening for MLD.

As you are aware, together we represent the clinical, laboratory and patient groups in the UK involved with Metachromatic leukodystrophy (MLD), a rare but devastating inherited form of childhood dementia. We remain extremely concerned by the recently published decision of the UK NSC to reject adding MLD to the UK newborn screening programme. Having had the opportunity to examine the UK NSC decision document in detail, we have taken the opportunity to highlight the key areas where we consider that the evidence review that supported the decision of the UK NSC remains subject to challenge.

I am sharing our summary paper identifying key areas where we suggest the UK NSC should take a further look at the evidence and the decision: 1) Test Accuracy (Criterion 4), Cut Off Levels (Criterion 5), Early Treatment Benefits (Criterion 9), Cost Effectiveness (Criterion 14), together with some more general additional points. We have also taken the opportunity to include proposals and recommendations.

We recognise that MLD is being considered as a possibility for inclusion in the EquipoISE project. Whilst welcoming this potential new development, we have deep concerns over the time it is going to take to get MLD onto the screening programme. Dr Graham Shortland presented the current position at the Specialised Healthcare Alliance (SCHA) meeting on 5th February 2025. Currently there appears to be no clear project plan, no defined timescales and no resources yet identified. This is the foundation for us pushing for an independent ISE for MLD.

We do recognise that you are fully aware of the importance of moving forward quickly with newborn screening for MLD. Since NICE approved the treatment in March 2022, 37 children have been diagnosed with MLD. 6 have fortunately received the treatment; however, 31 children have died or remain with a life limiting illness through late diagnosis and the unavailability of newborn screening in the UK.

In addition, clinicians, scientific experts and patient organisations are all very willing to assist the UK NSC and their project team in expediting the provision of evidence to ensure that newborn screening for MLD is added to the UK NBS programme at the earliest.

Yours sincerely,

Dr Arunabha Ghosh (Chair of the MLD NBS Steering Group)

On behalf of:

Lysosomal Storage Disorder Highly Specialised Service (HSS) Teams (paediatric)

- Great Ormond Street Hospital
- Birmingham Women's and Children's Hospital
- Manchester University NHS Foundation trust

NHSE Inherited White Matter Disease centres

Metabolic teams Scotland, Wales and Northern Ireland

Archangel MLD Trust

MLD Support UK

The MPS Society

LSD Collaborative

BIMDG

MetBioNet

UK newborn screening laboratory network (UKNSLN)

CC

Dr Graham Shortland, Vice Chair UK NSC

NHSE/ HSS Commissioners

NICE