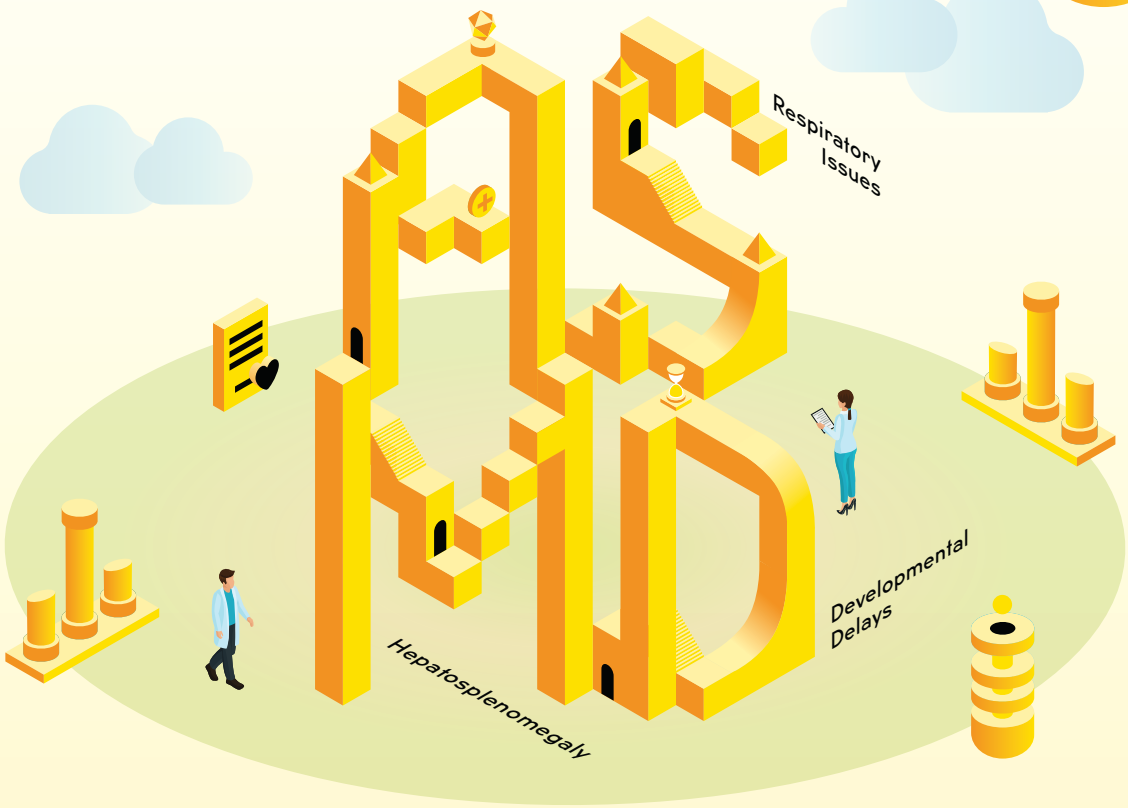
Miss the angle, **miss the diagnosis**



Change your perspective. **Change their journey.**

Acid Sphingomyelinase Deficiency (ASMD), commonly referred to as Niemann-Pick disease types A, B, and A/B, is a rare, progressive and often life-limiting lysosomal storage disorder.[1]

It's caused by a deficiency or absence of the enzyme acid sphingomyelinase, leading to the accumulation of sphingomyelin in vital organs including the liver, spleen, lungs and bone marrow.

ASMD spans a spectrum of severity:[1]

Type A (infantile neurovisceral): Rapidly progressive and fatal in early childhood

Type A/B (chronic neurovisceral): Intermediate form with mixed features

Type B (chronic visceral): Slower progression, but still with significant morbidity

ASMD symptoms mimic more common conditions and patients often face years of misdiagnosis.[1] HCPs however, can change that trajectory, by seeing familiar signs through a new lens.

Key symptoms may include:[1,2]

- Unexplained hepatosplenomegaly
- Thrombocytopenia or bleeding issues
- Recurrent respiratory infections or interstitial lung disease
- Short stature or developmental delays
- Family history of genetic conditions or consanguinity

Early recognition is vital. With increased awareness and timely investigation, patients and families can access care, support and clarity sooner.

What seems rare becomes recognisable.

Shift your perspective. **Change their journey.**

The ASMD Perspective Index* is a free, simple and engaging online index designed to support early identification. Developed with input from clinicians and patient groups, it helps guide decision-making when symptoms don't quite add up.

BEGIN YOUR JOURNEY WITH THE ASMD PERSPECTIVE INDEX*
DESIGNED TO HELP SHIFT YOUR PERSPECTIVE

Perspective reveals possibility.

How it works:

By taking this journey, you are able to:

- Select observed symptoms.
- Each selection builds the possible likelihood and reveals potential next steps.
- Once key symptoms are identified, the index prompts further consideration.

Your patient's path will be revealed:

- Based on the symptoms selected, the ASMD Perspective Index will guide you towards suggested next steps, including further monitoring, referral for enzyme assays and genetic testing, or guidance on tests to benchmark disease progression.

The symptoms remain the same; **it's your perspective that has shifted.**

References: 1. Wasserstein, M. et al. Molecular Genetics and Metabolism. 2019;126(2):98–105.
2. Doerr, A. et al. Molecular Genetics and Metabolism Reports. 2024l;38:101052.

*Please note: This ASMD Perspective Index is not intended for diagnostic purposes.

NPUK is here to help. We offer support for patients and families affected by ASMD, as well as the healthcare professionals involved in their care. Get in touch to access our services or download the clinician toolkit to learn more about ASMD.

Donate today at www.npuk.org Email: info@npuk.org Helpline: 0191 415 0693

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