Evelina London Children's Hospital Joins the INPDR Clinician Reported Database

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The International Niemann-Pick Disease Registry (INPDR) is proud to welcome Evelina London Children's Hospital in London, United Kingdom, as the latest site to join the Clinician Reported Database. This new partnership marks another important step in our mission to create a truly global resource for advancing the understanding, diagnosis, and treatment of Niemann-Pick diseases.

Led by Dr. Berna Seker Yilmaz, the site's Principal Investigator, the team at Evelina's will play a key role in expanding the reach of the Registry in the United Kingdom. Their participation will help ensure that patients and families in the region have the opportunity to contribute to a collective effort that benefits the global Niemann-Pick community.

By joining the INPDR, Evelina London Children's Hospital will support ongoing research and care improvements while empowering families to contribute their experiences and data to a global initiative driven by and for the Niemann-Pick community.

Shaun Bolton, INPDR Chief Operating Officer, who has been working closely with the team in London, commented: *"It's a pleasure to welcome Evelina London Children's Hospital and Dr. Seker Yilmaz to the INPDR. Their dedication to children with rare diseases and commitment to research will significantly strengthen our collaborative network. We are excited to collaborate with them to enhance the Registry and benefits the wider community."*

Dr. Berna Seker Yilmaz, Principal Investigator at Evelina London Children's Hospital, said: *"We are pleased to join the International Niemann-Pick Disease Registry and contribute to this important global initiative. Given the complexity and rarity of Niemann-Pick diseases, meaningful progress relies on strong international collaboration and the systematic sharing of knowledge. Through our participation in the INPDR, we aim to advance research and clinical understanding while ensuring that the experiences of patients and families in the UK are meaningfully represented in a collective effort to improve outcomes worldwide."*

We are honoured to collaborate with Evelina London Children's Hospital and look forward to working together to drive progress, raise awareness, and improve outcomes for those affected by Niemann-Pick diseases.

Notes to Editors

About Evelina London Children's Hospital

Evelina London is part of Guy's and St Thomas' NHS Foundation Trust. They're one of only two specialist children's hospitals in London. Their vision is to be a world leading centre of life-changing care for children, young people and their families.

They care for more than 104,000 children and young people each year at their hospital and community sites. Their neonatal unit cares for around 800 babies a year and has some of the best survival rates in the UK.

Learn more about Evelina London Children's Hospital here: Evelina London

About the INPDR

The INPDR is a web-based disease-specific registry, collecting information about ASMD Niemann-Pick Disease (types A & B), and Niemann-Pick Disease Type C, via, an anonymised Clinician Reported Database (CRD) and a Patient Reported Database (PRD). The PRD enables patients to self-enrol online and to contribute their data through a series of questionnaires including disease impact, health economics and quality of life. The INPDR is actively supported by patients, clinicians, patient advocates and researchers from over 20 countries across five continents.

For more information, visit: www.inpdr.org.

About Niemann-Pick diseases

Niemann-Pick diseases are divided into two distinct entities: (1) acid sphingomyelinase-deficient Niemann-Pick disease (ASM-deficient NPD) resulting from mutations in the SMPD1 gene and encompassing type A and type B as well as intermediate forms; (2) Niemann-Pick disease type C (NP-C) including also type D, resulting from mutations in either the NPC1 or the NPC2 gene. Both Niemann-Pick diseases have an autosomal recessive inheritance and are lysosomal lipid storage disorders, with visceral (type B) or neurovisceral manifestations.

Acid Sphingomyelinase Deficiency (ASMD; alternatively known as Niemann–Pick Disease Types A, B and A/B) is an ultra-rare multisystem genetic disorder caused by pathogenic variants of the SMPD1 gene. Clinical features, time of onset and disease severity can vary greatly among the subtypes and even within families bearing identical genetic alterations. At the severe end of the spectrum, the disease is rapidly progressive in nature and results in premature death. At the milder end, patients may be oligosymptomatic and a diagnosis can be easily overlooked. The rarity of the disease and the scarcity of expertise contribute to misdiagnosis, delayed diagnosis and barriers to adequate care. ASMD is a pan-ethnic ultra-rare, autosomal recessive metabolic disorder, with an estimated global prevalence of ~ 1:100,000–1,000,000 births.

Niemann-Pick Type C (NPC) is a progressive and life limiting autosomal recessive disorder caused by mutations in either the NPC1 or NPC2 gene. Mutations in these genes are associated with abnormal endosomal-lysosomal trafficking, resulting in the accumulation of multiple tissue specific lipids in the lysosomes. The clinical spectrum of NPC disease ranges from a neonatal rapidly progressive fatal disorder to an adult-onset chronic neurodegenerative disease. The age of onset of the first (beyond 3 months of life) neurological symptom may predict the severity of the disease and determines life expectancy. NPC has an estimated incidence of ~ 1: 100,000 and the rarity of the disease translate into misdiagnosis, delayed diagnosis and barriers to good care.