International Niemann – Pick Disease Registry

Announces Departure of Chief Executive Officer Conan Donnelly

18 March 2025 – Tyne & Wear, England.

The **International Niemann-Pick Disease Registry (INPDR)** announces that **Conan Donnelly will be stepping away from his role as Chief Executive Officer and Director of INPDR Gateway.** Since joining INPDR, Conan has been instrumental in driving the organisation's growth, strengthening collaborations, and expanding its reach within the global Niemann-Pick community.

Reflecting on his time at INPDR, Conan shared: "It has been the privilege of my life to help INPDR grow and evolve. The dedication of the board, our talented team, and our partners has made this journey incredibly fulfilling. I am deeply grateful for the trust, support, and collaboration from the entire community."

Under Conan's leadership, the INPDR has experienced significant growth, deepening relationships with key stakeholders across the Niemann-Pick community and reinforcing the Registry's role as a critical resource for research, clinical care, and advocacy. His contributions have laid a strong foundation for the Registry's continued success.

Jim Green, Chair of INPDR, expressed his gratitude for Conan's leadership: "Conan's impact on INPDR has been profound. His passion, vision, and dedication have been instrumental in advancing the Registry's work, ensuring that we continue to serve patients, families, clinicians, and researchers. We sincerely thank him for his unwavering commitment and wish him the very best in his future endeavours."

With the expansion of the Registry and the development of a new strategic plan, Conan's departure marks a natural point of transition. The INPDR's dedicated team and governance structure will ensure continuity and ongoing projects and partnerships will continue as planned. Further updates on the leadership transition will be shared in due course.

Notes to Editors

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.