International Niemann-Pick Disease Registry announces new research showing the impacts of Olipudase Alfa on adults with ASMD: The Patient-Reported Experience

11 March 2024 – Tyne & Wear, England. The International Niemann-Pick Disease Registry (INPDR) today announced a new research project to demonstrate the impacts of Olipudase Alfa on adults with ASMD through the Patient Reported Database. Acid sphingomyelinase deficiency (ASMD) is a rare recessive lysosomal disorder caused by a deficient activity of the acid sphingomyelinase (ASM) enzyme. Olipudase alfa, an enzyme replacement therapy, has recently been approved in several countries for the treatment of the non-neurologic manifestations of ASMD. While studies demonstrate clinical benefit, little is known about its impact on quality of life (QoL). To address this gap, researchers from the International Niemann-Pick Disease Registry in partnership with Niemann-Pick UK, the National Niemann-Pick Disease Foundation and the International Niemann-Pick Disease Alliance conducted a study to better understand the real-life experience of adults with ASMD and assess the impact of olipudase alfa on QoL.

The study involved issuing a questionnaire to adults (>18 years) with a confirmed diagnosis of ASMD who had received olipudase alfa as an experimental or approved treatment. Eleven adults were included in a final analysis and seven participants also took part in semi-structured interviews. Key study findings included extensive physical burden of the disease, with bone pain, abdominal pain, shortness of breath, nose bleeds, diarrhoea, abdominal cramps, fatigue and chronic headaches all frequently reported. Treatment with olipudase alfa was associated with reduced symptom prevalence and frequency, resulting in improved quality of life for participants. The perceived benefits of treatment outweighed any safety risks identified.

"These findings highlight a significant burden of disease for people with ASMD as well as the positive impact of olipudase alfa, both in terms of symptom improvement and quality of life. The study underscores the importance of olipudase alfa as a treatment option for individuals with ASMD, emphasizing the need for continued access to this therapy for all patients. " said Toni Mathieson, study author and Chief Executive of NPUK.

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-enroll 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 disease

Niemann-Pick diseases are a group of rare and devastating inherited lysosomal storage disorders that can affect both children and adults: Acid Sphingomyelinase Deficiency (ASMD) includes Niemann-Pick disease type A (NPA) and type B (NPB), which are caused by a lack of the enzyme acid sphingomyelinase leading to a build-up of toxic materials in the body. Niemann-Pick disease type C (NPC) is a hugely life-limiting neurodegenerative disease caused by an accumulation of lipids (fats) in the liver, brain, and spleen.