The International Niemann-Pick Disease Registry Welcomes Soroka Medical Centre as the First INPDR Site in Israel

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The International Niemann-Pick Disease Registry (INPDR) is proud to announce that Soroka Medical Centre in Beer Sheva, Israel, has officially joined as an INPDR site, integrating the Registry into its research efforts. This milestone underscores the critical role of global collaborations in advancing understanding and treatment for Niemann-Pick Diseases (NPD).

Under the leadership of Dr Orna Staretz-Chacham, The Cheryl & Haim Saban children's Hospital at Soroka Medical Centre is a renowned hub for rare disease research and clinical care in Israel. By adopting the INPDR as its NPD registry, Soroka Medical Centre will contribute real-world patient data to the global network, driving progress in diagnosis, treatment, and research efforts.

Dr Orna Staretz-Chacham emphasized the significance of this collaboration: "Integrating the INPDR into our work at Soroka Medical Centre is a major step forward for the Niemann-Pick community in Israel. This partnership ensures that our patient data actively contributes to research and innovation, improving outcomes not only for our local patients but for families affected worldwide."

This development highlights the vital role that each INPDR site plays in ensuring that patient data informs clinical research, care provision and treatment approaches... With Soroka Medical Centre now fully involved, the INPDR continues to expand its global reach, reinforcing the power of collective knowledge in rare disease research.

Shaun Bolton, Chief Operating Officer of INPDR, welcomed the new partnership, stating: "Every new INPDR site represents a step forward in Niemann-Pick diseases research. Soroka Medical Centre's expertise and commitment will provide invaluable contributions to the Registry, ensuring that clinicians and researchers have the necessary data to improve patient outcomes. This also brings an opportunity for patients in Israel to actively support meaningful research. We are excited to collaborate with their dedicated team and further strengthen the global network of registry centres."

The INPDR remains committed to expanding its partnerships with leading medical and research institutions worldwide, ensuring that patient data is utilised effectively to accelerate progress for Niemann-Pick Diseases.

Notes to Editors

About Soroka Medical Centre

Soroka Medical Center is the only major medical center in the Negev region and one of Israel's largest, most advanced, and most active hospitals. As a tertiary referral and trauma center, we provide care to more than one million people in a region that accounts for 60% of the country's total land area.
Soroka is a strategic asset of the State of Israel and the country's "medical iron dome" in routine times and during emergencies for civilians and IDF soldiers alike.

Since October 7, 2023, Soroka has been at the forefront of the effort to save the lives of civilians and soldiers. Soroka's staff has demonstrated unparalleled heroism. When the war broke out, our teams faced an unprecedented number of wounded, more than any other hospital in the world has ever had to treat at once. Once again, Soroka's strategic importance and the remarkable dedication of our medical teams were demonstrated with striking clarity.

Soroka is recognized for its excellent clinical care and leads the country in many areas, including genetics, obstetrics and gynecology, infection control, and emergency care and preparedness. Each year at Soroka, there are approximately 600,000 visits to our outpatient clinics, 270,000 visits to our Emergency Department (the busiest in the country), some 84,000 inpatient admissions, more than 32,000 surgical procedures, and more than 17,000 births.

In the next decade, Soroka's rapid growth in construction and infrastructure will continue, driven by innovative processes and the advancement of cutting-edge technological services.

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.