P-079 - BUILDING BRIDGES IN RARE METABOLIC DISEASES: THE INTERNATIONAL NIEMANN-PICK DISEASES REGISTRY (INPDR)

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BACKGROUND: Niemann-Pick diseases (NPD) are a group of rare inherited Lysosomal Storage Disorders (LSDs) that can affect both children and adults. Niemann-Pick type A/B is caused by acid sphingomyelinase deficiency (ASMD), presenting with both neurological and/or visceral finding with varying effects on other organs including the lungs. Niemann-Pick Type C disease (NPC) is a trafficking lipid disorder caused by two different genes NPC1 and NPC2, leading to an accumulation of non-sterified cholesterol and sphingolipids in the liver, brain and spleen

OBJECTIVES: To describe the International Niemann-Pick Disease Registry (INPDR), a disease-specific registry collating the global data of patients diagnosed with Acid Sphingomyelinase Deficiency (ASMD) and Niemann-Pick Type C (NPC) materials and methods: Clinical, biochemical, neuroradiological data are collected, including the following items: demographic characteristics, genetic profile, clinical manifestations, quality of life assessment, among others.

RESULTS: Patient registries can fulfil a number of roles, including collecting data regarding disease natural history data, post-marketing surveillance tool and patient quality-of-life register. The International Niemann-Pick Disease Registry (INPDR) contains two linked, but separate, databases, one holding clinician entered data and the other containing patient recorded outcomes (PRO). Data from 236 patients, entered from 6 countries is presently held within the Registry. 74% of the patients entered in the Registry so far have NPC and 26% are diagnosed with ASMD. Most of the patients with NPC (71%) and ASMD (74%) were diagnosed below 12 years of age; in contrast, 15% of ASMD patients and 10% of NPC patients had their diagnosis after 30 years of age.

CONCLUSIONS: INPDR is a single, rare disease-specific registry collating Niemann-Pick data on a global basis. It was created by professionals and patients for worldwide use. It collects clinical data and patient reported data with separate datasets for ASMD and NP-C. It will replace the need for multiple registries and offer a single, effective data resource for NPD.