

The INPDR is owned by the **International Niemann-Pick Disease Alliance (www.inpda.org)**, an alliance of non-profit NP patient support organisations, developed and managed in collaboration with University Hospitals Birmingham and international experts.



This collaboration between clinicians, scientists, researchers, pharmaceutical companies and patient associations across the world, will collect clinical, genetic, diagnostic and outcome data from patients with Niemann-Pick Diseases (NPD).

This project has received funding from the European Union, in the framework of the Health Programme.

The INPDR aims to improve the care of patients with Niemann-Pick Disease (NPD) across the world, by:

- ⇒ **Sharing information** - collating anonymised patient data from across the world in a consistent format
- ⇒ **Gaining knowledge and insight** - establishing the natural history of NP diseases
- ⇒ **Improving research** - coordinating research efforts globally
- ⇒ **Enabling rapid diagnostics** - facilitating equitable access to state of the art diagnostic tests



To find out more, please visit our website

www.inpdr.org

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International Niemann-Pick Disease Registry

(INPDR)

A new concept in rare disease registries

Improving the care of Niemann-Pick patients across the world

www.inpdr.org



Co-funded by
the Health Programme
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Niemann-Pick diseases (NPD) are a group of rare inherited Lysosomal Storage Disorders (LSDs) that can affect both children and adults.



Niemann-Pick Type C disease (NP-C) is a rare, inherited and life-limiting metabolic condition, caused by an accumulation of lipids (fats) in the liver, brain and spleen. Without treatment, the vast majority of children with NP-C die before age 20, however more mildly affected patients can remain symptom-free until adulthood.

Accumulation of the lipids in the brain causes progressive neurological damage, loss of motor skills and mobility, dysphagia (problems with swallowing), seizures and dementia, and is associated with a poor prognosis.

Acid Sphingomyelinase Deficiency (ASMD) or Niemann-Pick Disease Type A and Type B are caused by a lack of Acid Sphingomyelinase (ASM). ASM is an enzyme that breaks down sphingomyelin, which has a functional and structural role in cells. However, it can be toxic if it builds up, causing cells to die and organs to stop working properly.

ASMD Type A is a rapidly progressing neurodegenerative disease which results in premature death usually before the age of 5.

ASMD Type B is a gradually progressing, variable condition resulting in enlarged liver and spleen with varying effects on other organs including the lungs.



Treatment of NPD

At present, there is no cure for NPD, although those affected may benefit from medicine to treat the symptoms of the disease. The ultimate aim of the INPDR is to encourage and enable research which will lead to effective

therapeutic options for this group of diseases.

Why is the INPDR needed?

A registry is an important tool for collecting and recording information about a disease: how it affects and progresses in an individual patient, and how it presents to those involved in diagnosis, treatment and research. Registries are typically held by the pharmaceutical industry to investigate a single drug. This can lead to several registries for the same disease, and limited access to the data gathered.

The INPDR is a disease registry owned by patient groups and clinicians involved in the care of NPD



patients. It will enable progress by allowing authorised access to anonymised clinical data, helping to identify and recruit patients to clinical trials and coordinate research efforts globally.

This will stimulate a step change in the volume and quality of research into NPD which will help to improve patient outcomes.