The International Niemann-Pick Disease Registry (INPDR)

A new model of patient-empowered data ownership and management

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The International Niemann-Pick Disease Registry (INPDR) has been developed by a consortium of professionals as part of an EU funded project and has been running live since October 2016. The Registry is owned and managed by the INPDR, a subsidiary of the International Niemann-Pick Disease Alliance (INPDA). The key aims of the Registry are to share information by collecting international anonymised patient data in a consistent format; to define the natural history of Acid Sphingomyelinase Deficiency (ASMD) and Niemann Pick Type C (NPC); to improve analysis by coordinating research efforts globally and to enable rapid diagnosis by facilitating equitable access to state of the art diagnostic tests. The Registry 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. The INPDA has membership from 15 countries and a major next step is to increase the amount of data held in the Registry and specifically encourage more countries to enter their clinical/patient data on to the Registry. An experienced Recruitment Consultant with extensive knowledge of Niemann-Pick disease is leading this exercise on behalf of INPDA. Her role will also include contacting potential clinician contributors, to help facilitate the entering of data and where possible, identify and overcome any barriers to data entry. She will also attend family conferences to help raise awareness of the PRO process and review data entered so far, to verify the appropriateness and accuracy.

International Niemann-Pick Disease Alliance

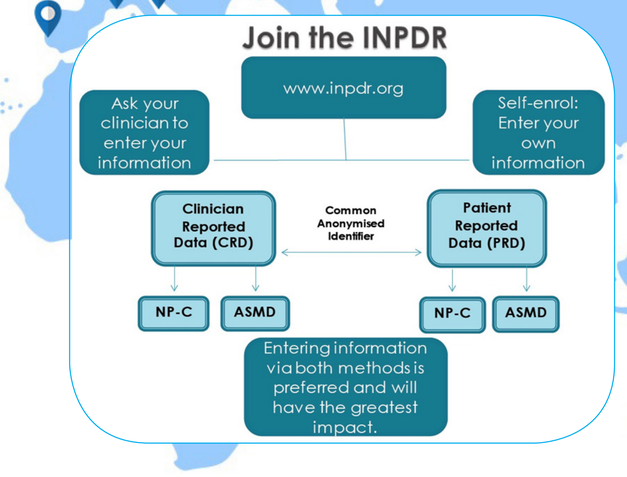
Niemann-Pick Disease (Overview)

- NPD are a group of rare inherited Lysosomal Storage Disorders (LSDs) that can affect both children and adults.
- ASMD Type A is a rapidly progressive neuro-degenerative disease which results in premature death usually before the age of 5.
- ASMD Type B is a gradually progressive neurodegen-erative condition resulting in enlarged liver and spleen with varying effects on other organs including the lungs.
- 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, although more mildly affected patients can remain symptom free until adulthood.
- All types of Niemann-Pick disease are acquired through autosomal recessive inheritance, where both parents are carriers of the faulty gene. Parents seldom know that they carry the disease and hence have no control over the transmission to their child.

INPDR

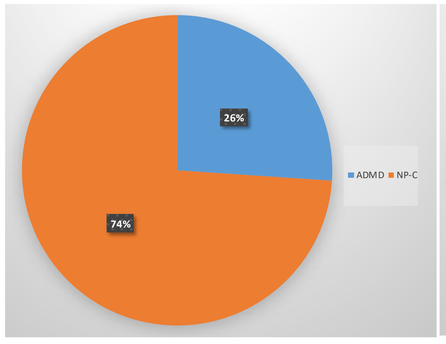
The INPDR is a not for profit, registered company based in the UK. It is a subsidiary of the INPDA (www.inpda.org) and has an independent Board of Directors. This initiative encourages global collaboration between patient groups, clinicians, scientists and researchers, all of whom wish to improve care and treatment options for NPD patients everywhere:

- The INPDR is a single, rare disease-specific registry collating Niemann-Pick data on a global basis:
- Created by professionals and patient groups for world-wide use with the support of an EU grant
- Uses the power of data to improve the understanding of NPD
- Supports and facilitates research and therapy development
- Replaces the need for multiple registries; the INPDR offers a single, effective data resource for NPD
- Encourages earlier diagnosis and equitable access to diagnostic testing
- A unifying initiative much-needed by patients and professionals alike

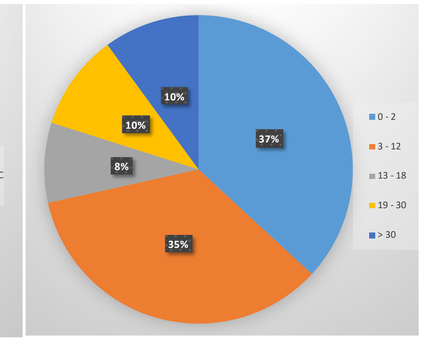


Snapshots of Registry Data (Examples)

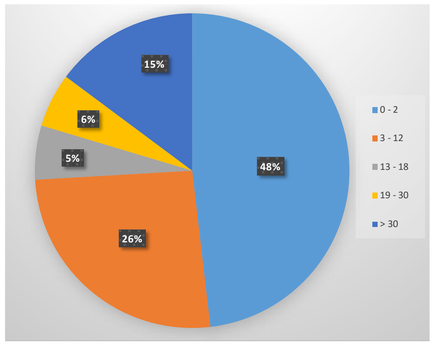
NP Type



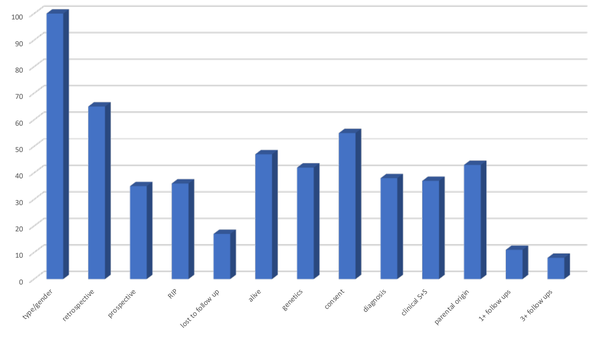
NP-C: Age of Diagnosis



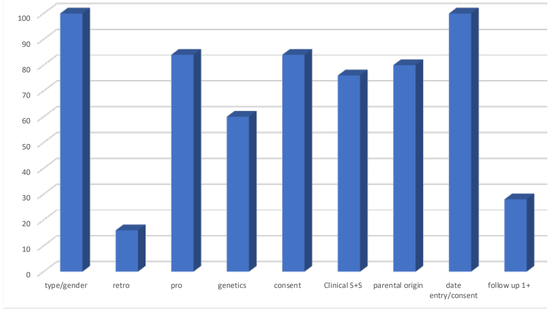
Type A/B: Age of Diagnosis



Percentage of live Type C patients with completed data



Percentage of live ASMD patients with completed data



KEY CHALLENGES

Ethics and approvals process varies in all countries

Clinicians and patients struggle to find time to enter data

Registry contains data from only 6 countries so far

Entering of follow up data is sporadic

Lack of awareness of how Registry operates

ACTIONS

Enrol more patients

Completeness/Accuracy

Complete thorough audit

Ongoing analysis of Registry

Review support structures

DESIRED OUTCOMES

- Encourage community engagement via robust recruitment strategy
- Extend Global membership
- Engage clinicians to enter data after every patient visit/contact
- Consider variety of methods and processes for entering data
- Identify what works well and what can be improved
- Redevelop and enhance the Registry during 2018
- Understand the impact of NPD on patients and families
- Improve understanding of NPD
- Enhance training and support material
- Establish a key contact in every member country

Acknowledgements

The INPDR would like to recognise the contribution of the global Niemann-Pick community, including those affected, their families and health/research professionals, whose on-going support of the Registry has enabled the creation of this valuable resource, which aims to enhance diagnosis, care and treatment for all affected and to encourage further research and understanding.