

International Niemann-Pick Disease Registry (INPDR)

A BEACON FOR RARE DISEASES

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BACKGROUND:

The International Niemann-Pick Disease Registry (INPDR) was created by a collaboration of patient groups, health and research professionals working in the field of Niemann-Pick diseases.

Niemann-Pick diseases are a group of rare genetic diseases with a distinct but overlapping clinical spectrum that ranges from a neonatal fatal disorder to an adult onset, chronic debilitating neurodegenerative disease. The diseases are pan-ethnic and patients are distributed throughout the world.

METHODS

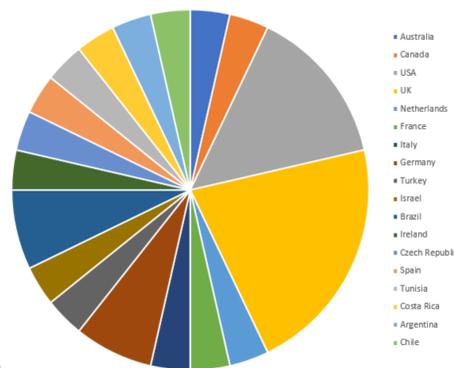
The study of rare diseases is typically characterised by a small and potentially disparate community of patients.

The active participation of clinicians and patients from around the world will provide robust data on this rare disease, facilitating the development and recruitment of future clinical studies and supporting access to new and emerging therapies.

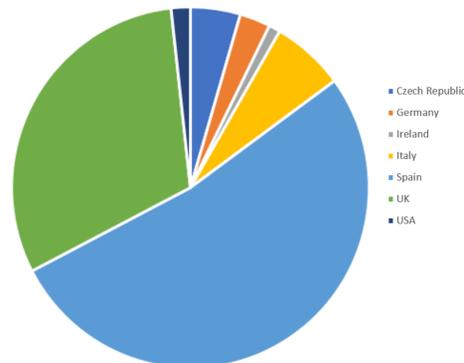
The INPDR recognises the importance of the patient perspective in understanding and assessing disease impact by combining the clinical and 'real-life' experience of patients to offer a much more powerful and meaningful source of research data.

- Includes both clinical and patient reported data for Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick Disease and Niemann-Pick Disease type C
- 28 centres from 18 countries over 5 continents are entering data or undertaking the R&D submission process
- Patient Reported Data (PRD) has been entered in 9 countries; allowing patients to self-enrol and contribute to progress, independent of clinician input
- Clinician Reported Data (CRD) has been entered in 7 countries
- The INPDR has been operational since 2014; it is an established, regulatory compliant registry with appropriate security, management and controls

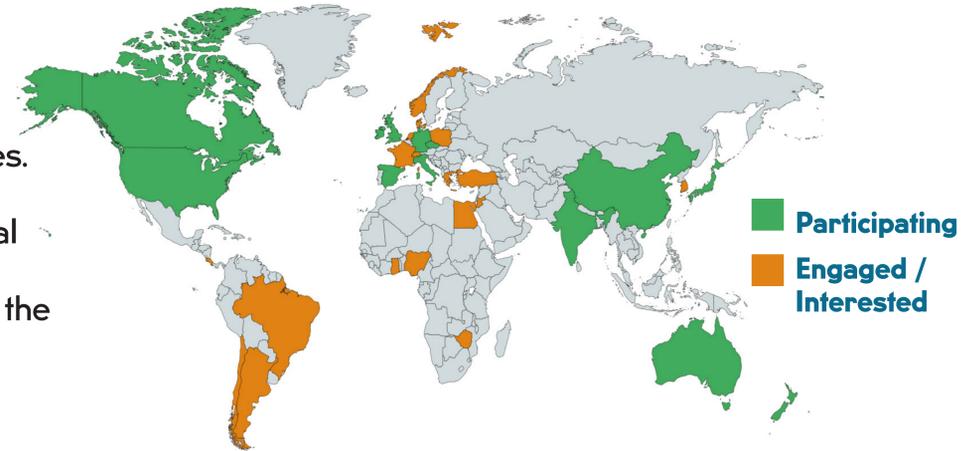
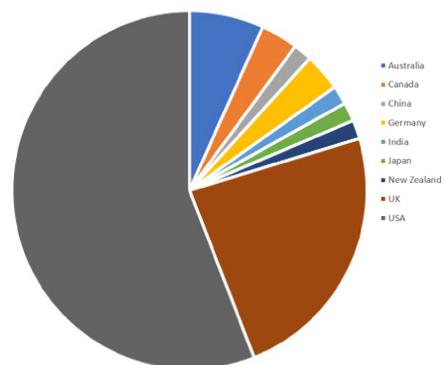
Countries entering data or involved in the process of R&D submission



CRD Patient entries per country at end of 2019



PRD Patient entries by country at end of 2019



VISION

The INPDR aims to improve standards of diagnosis, care and treatment for the global Niemann-Pick patient community:

- Aims to establish the natural history of ASMD NPD and NP-C
- Supports and empowers the patient community, clinicians, researchers and scientists by building stronger networks and providing access to up to date expert information, advice and signposting
- Moves away from the established norm of rare disease registries; those that focus on a single drug, not a disease
- Offers a single data resource to facilitate basic and clinical research and aid recruitment to interventional studies
- Supports interaction between national/supranational medicines agencies and industry

GOVERNANCE

Governance is overseen by a Board of Trustees, with advice and guidance provided by a Scientific Advisory Committee, comprised of clinical and research professionals, plus patient advocacy representatives.

DISCUSSION

The INPDR is undergoing a qualification opinion procedure of the Registry, with the European Medicines Agency (EMA), to ensure it is fit-for-purpose to perform pharmacoepidemiological studies and fulfil post-marketing activities for regulatory purposes in medicines indicated for the treatment of Niemann-Pick diseases. As such, INPDR offers a model for other rare diseases to accelerate understanding and medicines in their respective field.

CONCLUSION

The INPDR is a registry model that will help to inform and influence the wider rare disease community. Developed with patients at its heart, it has the potential to bring change and improvement to health care and services for those affected by Niemann-Pick and other rare diseases.

"...the INPDR offers the opportunity to collect information in a central location regarding all aspects of Niemann-Pick enabling a better understanding of the natural history of this rare disease..."



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