INPDR Forum 2025 – Meeting Report

Date: Saturday 20th September 2025
Location: Puerto Iguazu, Argentina
Time: 9:00am – 12:15pm
Facilitator: Solomon Mbua, INPDR Clinical Research Associate

Executive Summary

The 2025 International Niemann-Pick Disease Registry (INPDR) Forum convened in Puerto Iguazu, Argentina, bringing together over 60 participants including patient representatives, clinicians, researchers, and industry partners across 18 countries. Taking place every two years, the Forum's primary aim is to provide a comprehensive progress report, share research updates, and gather community and stakeholder input to help shape the future direction of the INPDR.

This year, key discussions focused on registry growth, data quality, and the importance of multi-stakeholder collaboration. Breakout sessions identified priorities, including improved communication, streamlined data entry, expanded language support, and enhanced feedback mechanisms.

1. Welcome and Opening Remarks

Speaker: *Prof. Tarekegn Hiwot, Chief Medical Officer*

Professor Hiwot opened the Forum by reflecting on the 14-year journey of the INPDR. He emphasised the importance of global collaboration in rare disease research, highlighting that no single country alone can collect sufficient data to understand the natural history of Niemann-Pick diseases.

He recalled the registry's origins as a patient-driven initiative born from a vision shared in Spain in 2012 and now recognised as a model of international partnership and empowerment. Professor Hiwot urged all attendees - families, clinicians, and industry partners - to continue contributing to the INPDR as a symbol of unity and hope for the Niemann- Pick

2. Keynote Speech: Value of the INPDR to All

Speakers: *Shaun Bolton (Chief Operating Officer) and Dr. Simon Day (Board of Trustee) – pre-recorded*

Shaun Bolton outlined the registry's mission to unify fragmented data across the Niemann-Pick field, enabling faster and more meaningful research. He explained the two data streams - clinician-reported and patient-reported - that together provide a holistic picture of the disease.

Shaun outlined the value INPDR can provide to groups working within Niemann-Pick diseases, including patients and families, clinicians, researchers, regulators and pharmaceutical companies. He also showcased INPDR's achievements in progressing research, collaborations and regulatory recognition, as well as plans to further the value of the INPDR.

Dr. Day complemented this with an industry and regulatory perspective, underlining the importance of standardised, high-quality data to answer critical questions about disease progression, treatments, and outcomes. He referenced ongoing studies on seizure burden, quality of life, and genotype-phenotype correlations, made possible through the registry.

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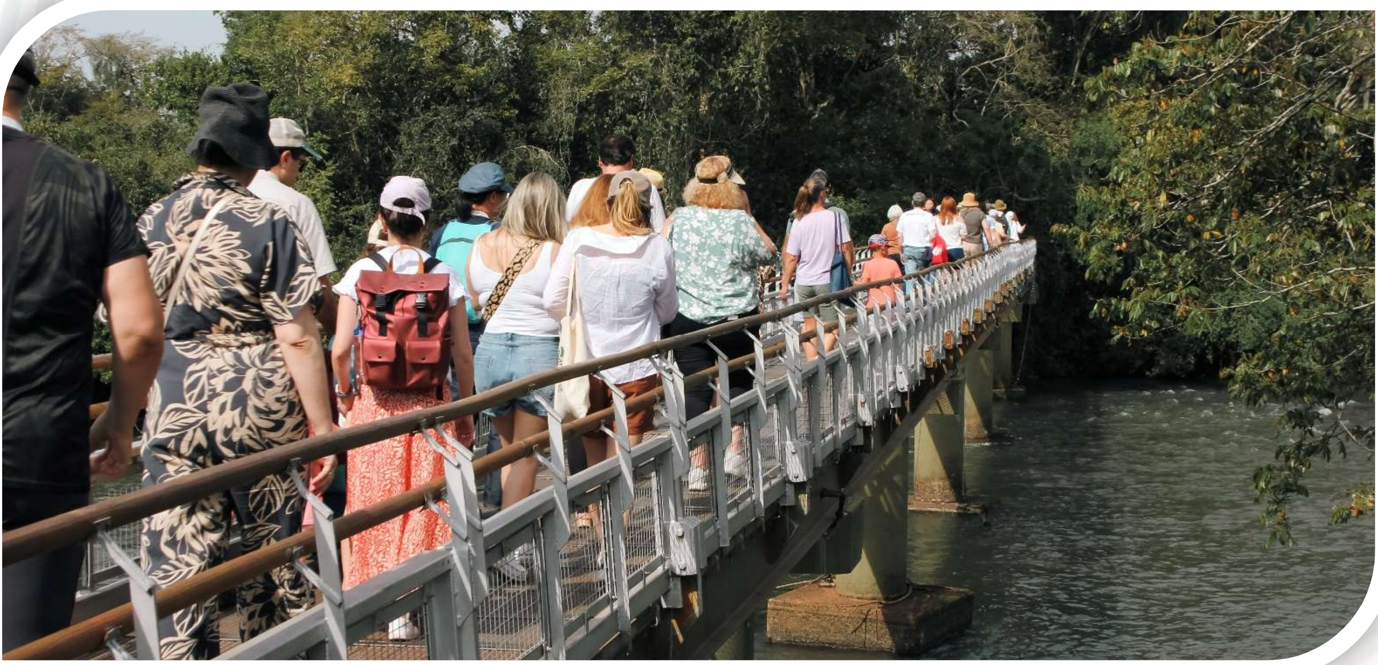

Both speakers closed by reaffirming the registry's guiding principle: *"By the community, for the community"*.

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3. INPDR Overview & Progress

Speaker: *Solomon Mbua, Clinical Research Associate*

Solomon provided a comprehensive update on the registry's growth, impact, and future goals. Since the last INPDR Forum which took place during the 2023 INPDA conference, 69 new patients have been added to the clinician-reported database, bringing the total to 502 across 29 active sites in multiple countries. The patient-reported database now includes 163 participants.

He highlighted achievements such as:

- Registry data used to support research projects, including 10 data access requests
- Development of the Global Unique Identifier (GUID) tool

- Publication of peer-reviewed research using registry data
- Securing funding from rare disease grant funders and industry engagement

Solomon outlined strategic priorities for 2026, including:

- Expanding global site participation
- Translating the patient database into six languages
- Developing reimbursement models for participating sites

He closed by thanking all contributors for their continued collaboration, reinforcing that collective effort drives the registry's success.

4. Research Update

Speaker: *Dr. Justin Hopkin, Chief Scientific Officer*

Dr. Hopkin shared an overview of recent and ongoing research supported by the INPDR, describing the registry as a "garden that is now bearing fruit."

He emphasised that the registry provides essential real-world data that accelerates study design and regulatory engagement and outlined how the INPDR can facilitate high-quality research

Ongoing projects include natural history studies, quality of life analyses, and clinical guideline development.

Justin encouraged continuous data entry by both clinicians and families, stressing that sustained participation ensures the registry remains a cornerstone for rare disease research and a model for other patient communities.

5. Perspectives: Community, Clinician, and Industry

This session brought together voices from three key stakeholder groups:

- Community Perspective – Mandy Whitechurch (Australian NPC Disease Foundation, Australia) shared how families' data contributions directly inform clinical care and research. She encouraged advocacy groups to promote registry engagement and highlighted the value of patient surveys.
- Clinician Perspective – Dr. Caroline Hastings (UCSF Benioff Children's Hospital, USA) explained how the registry supports global clinical collaboration, informs patient management, and strengthens long-term care strategies.

- Industry Perspective – Dr. Stefan Kolb (Switzerland) outlined how registries like the INPDR enables efficient trial design, reduce costs, and enhance regulatory compliance. He commended the INPDR for its independence and data quality.

Together, these perspectives reaffirmed the registry's central role in uniting all sectors to improve outcomes for Niemann-Pick patients worldwide.

6. Breakout Sessions

Facilitators: *Solomon Mbua and Table Leads*

The breakout discussions gathered input from clinicians, families, researchers, and industry representatives on how to enhance the INPDR's functionality, reach, and impact.

Key Themes Identified:

1. Transparency & Communication: Need for clearer communication about processes, timelines, and benefits.
2. Simplification: Streamlining forms and reducing duplication with national registries.
3. Language & Accessibility: Expanding translations and using simple, inclusive language.
4. Engagement: Strengthening clinician and patient participation, supported by advocacy groups.
5. Feedback: Creating feedback loops so contributors see how their data drives progress.
6. Technology: Exploring mobile apps and short educational videos.
7. Ethics & Consent: Integrating consent at diagnosis or clinical trial entry.
8. Local Adaptation: Supporting national requirements and community awareness efforts.

9. Collaboration: Establishing an ongoing working group for continuous improvement.
10. Data Access: Increasing transparency in data access fees and permissions.

The discussion concluded with a commitment from the INPDR team to review and implement actionable recommendations, ensuring the registry evolves in line with community needs

Wrap-Up

The Forum closed with a summary of outcomes and next steps. Participants agreed that the INPDR's mission remains strong, but its value must be communicated more effectively to clinicians, families, and other stakeholders worldwide.

Solomon thanked all attendees for their contributions, reaffirming that the insights shared during the Forum will guide future improvements and help the registry remain a global, community-driven platform for advancing research into Niemann-Pick diseases.

The INPDR will strive to provide regular updates on the progress of work identified through the Forum, including during future conferences and community updates.

For more information, please contact the INPDR via email at info@inpdr.org or visit our website at www.inpdr.org.