INPDR Expands in South America with New Partnership at Casa Dos Raros in Brazil

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The International Niemann-Pick Disease Registry (INPDR) is pleased to announce its latest partnership, welcoming Casa Dos Raros in Porto Alegre, Brazil, as a new Registry site. The second Registry site in Brazil, this development further strengthens the global effort to improve the understanding, diagnosis, and treatment of Niemann-Pick diseases.

By establishing a deeper presence in Brazil, the opportunity for Brazilian patients living with Niemann-Pick Diseases to share their experiences and contribute valuable data to advance the global understanding of Niemann-Pick diseases has increased, in addition to expanding the INPDR's global footprint.

With the support and national leadership of the patients, families, and clinicians of Brazil, together, we will continue to grow the Registry throughout the country. The data collected will play a vital role in research, patient care, and disease awareness—both locally and globally.

Professor Roberto Giugliani, Chief Investigator of INPDR coordinating center in Brazil, shared: "This is a major move for INPDR in Brazil, as Casa dos Raros has the necessary infrastructure and the skilled clinical research team to expand the reach of the registry in our country".

Dr. Carolina Fischinger, Principal Investigator at the Casa dos Raros' site, mentioned that "to have a site of INPDR at Casa dos Raros will enable the collection of data from all over the country, due to the countrywide presence of our institution and to its reputation in the Brazilian LSD community"

Solomon Mbua, INPDR CRA Consultant, who has been working closely with the team in Brazil, stated that "it has been a privilege to support Casa Dos Raros in joining the INPDR CRD. Their dedication to the rare disease community and commitment to clinical research will make a meaningful impact. We look forward to seeing the contributions they bring to the database and the broader Niemann-Pick community."

Notes to Editors

About Casa Dos Raros

The Comprehensive Care and Training Center for Rare Diseases was built in Porto Alegre through a partnership between the Genetics for All Institute and Casa Hunter, two civil society organizations that develop projects in the field of rare genetic diseases.

As a pioneering initiative in Latin America, Casa dos Raros was created with the purpose of establishing an interconnected network for comprehensive care for people with rare diseases. The goal is to provide integrated and multidisciplinary support to patients and families through rapid and accurate diagnosis, advanced treatments, clinical research focused on rare genetic diseases, and the training and qualification of healthcare professionals working in this field.

Learn more about Casa Dos Raros here: Casa dos Raros

About the

International Niemann

Pick Disease Registry

(INPDR)

The INPDR is a global non-profit organisation which has developed a web-based disease-specific registry, collecting information about ASMD Niemann- Pick Disease (types A & B), and Niemann-Pick Disease Type C, via, an anonymised Clinician Reported Database (CRD) and a Patient Reported Database (PRD). The PRD enables patients to self-enrol online and to contribute their data through a series of questionnaires including disease impact, health economics and quality of life. The INPDR is actively supported by patients, clinicians, patient advocates and researchers from over 20 countries across five continents.

For more information, visit: www.inpdr.org.

About Niemann-Pick diseases

Niemann-Pick Diseases are a group of rare, inherited disorders, affecting both children and adults. They happen because the body can't properly break down certain fats. These diseases are split into two distinct subgroups: Acid Sphingomyelinase Deficiency (ASMD) and Niemann-Pick Disease Type C (NPC).

Acid Sphingomyelinase Deficiency (ASMD) is an extremely rare, progressive genetic disease, previously known as Niemann-Pick Disease Type A (NPA), Type A/B and Type B (NPB). ASMD represents a spectrum of disease, caused by a lack of the enzyme acid sphingomyelinase and resulting in potentially life-limiting illness in children and young adults. NPC is a cellular lipid trafficking disorder, is a significantly life-limiting neurodegenerative disease that can present in different ways. NPC is progressive, with disabling neurological symptoms.