February 2023 Newsletter

Today is Rare Disease Day. Gaucher patients and their caregivers worldwide are showing their colours in support of this important day.

Find out how you can take part.

One of the ways the IGA marked Rare Disease Day was by taking part in a tweet chat with @DataSaves_Lives, @INPDR_registry and @eurordis to

discuss how real world data collected via patient registries, like **GARDIAN**, can ultimately transform lives.

Read the conversation

The IGA showcases GARDIAN at WORLD

IGA honorary President and International GARDIAN Limited Chair Jeremy Manuel, left, reports on the WORLD symposium held on the 22nd to 26th February in Orlando Florida.

Having just returned from the WORLD (**W**e're **O**rganising **R**esearch on **L**ysosomal **D**isease) Symposium I wanted to share with IGA members and newsletter readers my reflections on this energetic, exciting, and inspirational meeting.

The organisers describe WORLD as an international multidisciplinary forum for presenting and discussing the latest advances in basic science, translational studies and clinical trials on lysosomes and lysosomal diseases. It is all this and a lot more with formal sessions beginning at 6.15 am and running until 5 pm with meetings and dinners and panel discussions going on to the late evening. The lectures, with experts presenting on their research, the opportunities for the exchange of ideas, the updates and panels certainly serve

to encourage collaboration and the pursuit of further understanding. In addition to formal lectures and discussions there were poster sessions where research, discoveries and the latest developments were presented on electronic posters and where the authors explain to attendees the detail of their studies and findings.

The IGA hosted a booth which I “personed” together with Aviva Rosenburg, IGA Vice Chair, which had leaflets about the GARDIAN Registry. The booth was well visited, and the GARDIAN Registry created much interest, as did the poster accepted by the organisers for display entitled “A Collaborative approach to address the unmet needs of patients with neuronopathic Gaucher Disease Type 2 and Type 3, the creation of GARDIAN a patient-led and patient- owned global registry”. Dr Elin Haf Davis of Aparito who, together with IGA and IGL Chief Executive Tanya is one of the posters authors, and I stood by the poster explaining the way that the GARDIAN registry works.

There were approximately 2000 attendees at WORLD, with many representatives from the pharmaceutical industry, many (the larger ones) with their own booths, displays as well as representatives from start up companies looking to developing new therapies including Gene therapy. These symposia always provide the opportunity for patient groups to engage with scientists and clinicians, and with representatives from industry, and I was happy to update clinicians and companies on many aspects of the work of the IGA and the progress made and the plans for developing the GARDIAN Registry. I met with many companies, and presented at a patient panel and I am pleased to report that the work of the IGA and my description of the GARDIAN Registry was enthusiastically received.

In addition, I participated in a reception to launch the Global LSD collaborative, a new group of which the IGA is an important part, designed to bring together the various LSD groups to work together to address their common aims.

I left the WORLD Symposium having learned a great deal, having taken the opportunity of meeting many existing contacts and collaborators and making many new ones for the IGA and IGL. I also left greatly encouraged by all the activity that is going on that will undoubtedly benefit the Global Gaucher Community.

Charitable Access Success

Working with Sanofi, the IGA has supported a patient in Uganda to get access to treatment though the International Cerezyme Access programme.

Visit our website for more information on Charitable Access.

Volunteer Spotlight

Patricia Lucki

Regional Manager, Central America 1

I decided to volunteer as Regional Manager for the IGA because I believe in the need for a more sustainable world and I cannot understand sustainability without solidarity. The Regional Manager programme allows organized Gaucher patients to reach those who have no local support or framework and this adds meaning to our own local work. I expect to reach, hand in hand with the IGA, underserved patients and create a network in the Central American Integration System (SICA) region.

In practical terms, this means to include in the regional health agenda rare diseases research, treatment and care and to have a comprehensive catalogue that includes Gaucher disease in the regional diseases classification; to improve regional registries; and to have clear policies regarding treatment acquisition and registration. This year we expect to start a virtual reference center and this will be the first step towards our final objectives.

Please contact Vesna if you are interested in volunteering with us.

Type 1 Gaucher trial

Recruitment is open for a study to evaluate the safety and efficacy of Prevail's PR001 experimental gene therapy for Gaucher disease Type 1 (GD1).

The study is open to patients aged 18 to 50 years who have been on ERT or SRT for at least two years. For each patient the study will be approximately 5 years in duration, including up to a 45-day screening period. Patients will be evaluated during the first 18 months after dosing and will be followed up for an additional 42 months to monitor safety, immunogenicity, and selected biomarker and efficacy parameters.

Patients are being sought at the Lysosomal & Rare Disorders Research and Treatment Center in Fairfax, Virginia, U.S.

For more information visit bit.ly/prevailtrial

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