June 2023 Newsletter

20-21 April, London, UK

The 3rd International Conference on Lysosomal Diseases

This conference looked at recent advances in the understanding, diagnosis and treatment of Lysosomal Storage Disorders (LSDs) at large, but with a particular focus on the main (relatively more-common) Lysosomal Diseases: Gaucher, Fabry, Pompe and the various forms of MPS, and brought together international leaders from basic science to academic medicine, as well as representatives of the industry who take part in the development of innovative methodologies that will lead to new therapeutic modalities.

At this year's conference four new sessions dedicated to controversies in LSDs were introduced, including issues related to newborn screening, PGD-M (preimplantation genetic testing for monogenic disorders) for the prevention of mild LSDs, importance of patients' reported outcome measures (PROm) and the challenges of recruiting patients to clinical trials of gene therapy.

IGA CEO, Tanya Collin Histed writes: "I was invited by Prof Ari Zimran to participate in the conference and joined Prof Derralynn Hughes in the controversy session on 'the challenges of recruiting patients to clinical trials of gene therapy'. The 45-minute session saw Prof Hughes present on 'Very Important' and I presented 'Very Concerning'. It is very important and I believe healthy to be able to have these types of open and public discussions on critical topics for our community. It enables the probing and challenging questions to be asked to ensure that when making serious and life-changing decisions all the information is on the table and available to the patient and/or caregiver when faced with the choice of participating in a clinicial trial.

The conference programme included talks from Prof Timothy Cox, Prof Hans Aerts, Dr Robin Ely, Prof Jim Shayman, Dr Aimee Donald, Prof Arndt Rolfs, Dr Andrea Dardis, Prof Pilar Giraldo and many more. The programme can be found: Congress Program - Med-Lysosomal2023

ONLINE SURVEY

Should rare diseases be screened at birth?

EURORDIS, in conjunction with Screen4Care, is conducting a survey on newborn screening for rare diseases, and the IGA is encouraging Gaucher patients and their carers to take part.

The survey is an opportunity to share what you think are the possible benefits and detriments related to newborn screening of rare diseases: for example, in terms of anxiety, access to care or adjustments to family life.

This survey will take around 20 minutes to complete. It will help EURORDIS-Rare Diseases Europe, a non-profit alliance of 1000+ patient organisations, including the IGA, to work towards and advocate for a better use of newborn screening.

The more Gaucher patients and caregivers that answer the survey, the more relevant the results will be for the Gaucher community, so we encourage you to take part.

The survey is secure and the results are anonymised.

For more information and a link to the survey, visit https://bit.ly/newbornscreeningsurvey

Take the survey

Report on the First Romanian Conference of Patients with Lysosomal Diseases


Between May 23-24, 2023, the First Conference of Patients with Lysosomal Diseases from Romania was held in Cluj-Napoca, with "A Voice for the Future" theme. The

conference was organized by the Romanian Foundation for Lysosomal Diseases at the Grand Hotel Napoca from Cluj-Napoca. 100 people, patients, participated with lysosomal diseases from all over the country, belonging to them, but also doctors who diagnose and treat such patients.

Some of the participants came for the first time to such an event, others were also present at the Romanians for Lysosomal Diseases (FRBL) Foundation's events organized in the past. FRBL has now organized for the first time an event dedicated to all people with lysosomal diseases, including not only patients with Gaucher disease, but also those with the Fabry, Pompe, Nieman-Pick disease, Mucopolysaccharidoses of type I, II, III and IV.

Read more

Trailer and all episodes

Volunteer Spotlight

Jasenka Wagner

Jasenka Wagner is type 1 Gaucher patient from Croatia. She is founder and chair of the Croatian Gaucher association and member of the board of the Croatian alliance for rare diseases. She is university professor of human genetics and clinical biochemistry at the Medical faculty in Osijek, Croatia.

Her main motivation for being involved as a volunteer for IGA is raising knowledge and awareness about Gaucher and other rare diseases, as well as improving patients care and quality of life worldwide.

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

IGA Gene Therapy Educational Programme working group

We need two volunteers to join this group. We are looking for patients, caregivers or medical students. Initially, you will be involved for one year attending online meetings of the working group, reviewing and commenting on the website development, and other tasks associated with the project. It won't take more than one hour per week.

If you are interested in gaining new knowledge and skills in the area of gene therapy and website development, you can communicate in English, and can communicate online, please apply for this volunteer position.

Contact Vesna at vesna@gaucheralliance.org and find out more about the project and how can you contribute.

GARDIAN UNLOCKS

Enhancing outcomes for those with neuronopathic Gaucher Disease

GARDIAN

GARDIAN DATA HELPS US

TO SHOW THE REAL GLOBAL PICTURE OF nGD THROUGH THE PERSPECTIVE OF CAREGIVERS AND PATIENTS

GARDIAN
is already showing results
and there is still time to play your part!
Join the GARDIAN Registry today at:
gardianregistry.org

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