May 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

Amsterdam May 2023

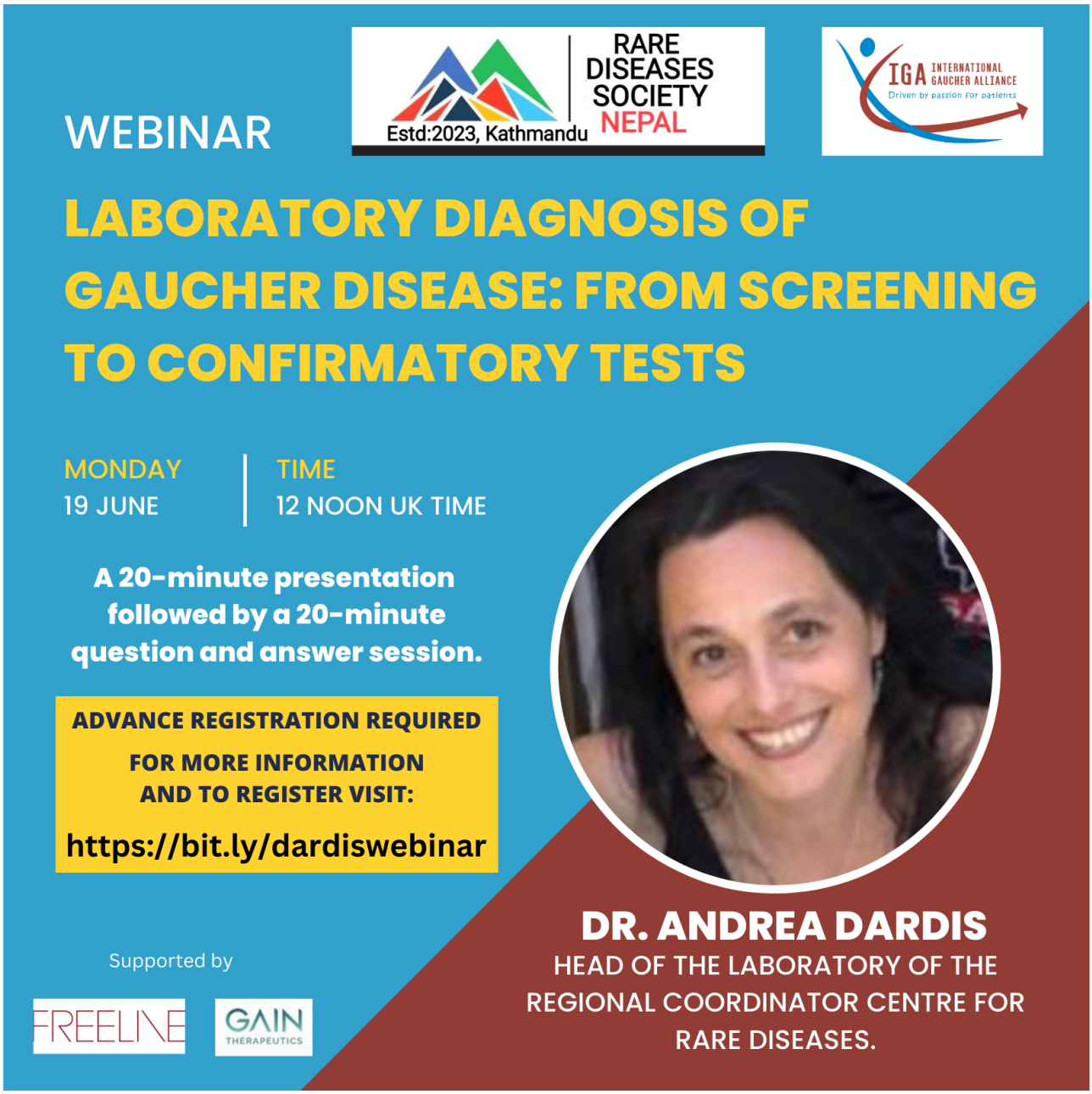
Gaucher Leadership Forum (GLF)

The GLF is a scientific meeting sponsored by Sanofi that brings together participants from around the world, including physicians, academics, and to discuss current care for patients with Gaucher disease and the direction of future disease management. CEO, Tanya Collin Histed writes:

Sandie Cowie, President of International Niemann-Pick Disease Alliance (INDPA) and I were invited to join the GLF at the end of day one of the two-day meeting to participate in a panel session moderated by Cristina Cardosa, Global Public Affairs Lead, Rare Nephrology and Cross Rare Initiatives from Sanofi.

The session titled “Gaucher Disease, an advanced but unfinished progress. Setting the path to accelerate Gaucher and ASMD patients’ journey” explored the progress made after 30 years of a first treatment for type 1 Gaucher disease and the remaining challenges, or unfinished business, and the vision and challenges for the ASMD community now that they have a licensed treatment. Topics included diagnosis, patient reported data, registries, and ended with Sandie and I addressing a message to the participants about what we see as key for our communities. This included collaboration, mental health, and wellbeing, not stopping with what we know but continuing to push boundaries to learn more so we can achieve better patient outcomes.

Patient advocates involvement in these types of meetings are vital, it is only through communication and collaboration that we can ensure that the patient is at the centre of all the discussions and work that is being done in their name.



Register now

LEAP2MONO Clinical trial for Type 3 Gaucher disease patients

The study is a 52-week study, opened to patients ≥12 and <18 years and adult patients with Gaucher disease Type 3 (GD3) who have been treated with Enzyme Replacement Therapy (ERT) for at least 3 years.

The study is to evaluate the efficacy and safety of **daily oral Venglustat versus intravenous Cerezyme** infusions every two weeks for improvement or stabilization of the neurological manifestations and maintenance of systemic disease stability. **Venglustat** is

an oral substrate reduction therapy that reduces the number of fatty substances in the cells and therefore helps to reduce their build up.

The study is double blind (meaning the patient and doctor will not know if they are receiving the study drug or not) and double dummy (the patient will receive the oral therapy which may or may not be the study drug) **and** an infusion (which may or may not be ERT).

The International Gaucher Alliance is planning on hosting two webinars about this clinical trial in early July 2023, more information will be posted on our social media platforms and sent to our members organisation, pre-registration will be required. More details, including how to register, will be posted on our social media and included in the next newsletter.

More information on this phase 3 clincial trial that opened in 2022

Gaucher patients in Sudan

The IGA is in touch with several Gaucher families, treating doctors and the pharmaceutical companies that provide charitable access treatment to the patients in Sudan. We are working to provide support and find ways that patients can continue to receive their infusions whether they have left or remained in Sudan.

If any needs our support, please reach out to us and we will try and help in any way we can. We hope that the situation in Sudan improves and that our Gaucher community can continue to receive the care and treatment they need and have worked so hard to get.

Volunteer Spotlight

Carine Alsokhn

Carine Alsokhn is a Gaucher Type 1 from Lebanon. She is a public health officer currently based in Geneva, Switzerland and has experience working with global agencies and collaborating with ministries of health and other relevant national bodies. She previously worked at the Ministry of Public Health in Beirut.

She also participated in many awareness campaigns and helped spread the word about Rare Diseases in general and Gaucher Disease in particular, in Lebanon and the region. She is a registered dietitian fluent in three languages Arabic, French and English.

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

Click here for more information and to register

International Gaucher Alliance

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