

The European Working Group on Gaucher Disease

Newsletter

Issue 1 October 2019

Dear Friends and Colleagues

It gives me great pleasure to bring you this first edition of the EWGGD newsletter to mark *International Gaucher Day* and the new work of the EWGGD. As many of you will be aware the 2019 meeting in Clermont Ferrand was a nidus for reinvigorating our organisations with a new determination to work together for the benefit of the whole Gaucher community. In this newsletter we will be introducing our new board, outlining our plans for promoting research and education in the area of Gaucher disease and our commitment to involve and communicate with all who have an interest in this important condition. I hope you enjoy reading this newsletter and will be enthusiastic about getting involved.

With best wishes to all

Derralynn Hughes

Chair European Working Group Gaucher Disease

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Mission statement

The aim of the EWGGD is to promote clinical and basic research into Gaucher disease for the ultimate purpose of improving the lives of patients with this disease; it brings together clinicians, scientists and patients in an open forum for discussion on all aspects of the condition.

The opportunity for presenting unpublished scientific data as well as free discussion is a central premise of the Group. For the dissemination of information, a two yearly workshop is organized. Although a limited number of representatives from industrial partners are invited to attend the meetings, the Working Group has always been an independent organization with a charitable purpose.

International Gaucher Day:



International Gaucher Day (IGD) first launched in 2014, by the EGA, based on an idea from participants at Go with Gaucher in 2012; a project working with young Gaucher patients. 'Rare but Not Alone' is the slogan of IGD. The aim of IGD is to raise awareness of Gaucher disease and to highlight the impact of Gaucher disease on the lives of children and adults. Throughout the 50 member countries of the IGA patients, families, doctors, scientists, healthcare professionals and the pharmaceutical companies will be marking IGD by holding events and activities. The IGA encourages its member patient groups to mark the day by organising a lecture for students at a medical school; arranging a lecture for doctors, nurses and clinicians at a hospital; gathering in a public place and spreading information about Gaucher disease; organising a workshop for journalists and doctors; inviting your own 'rare star' – to participate in the social media campaign; organising a sporting event: run, walk or bike ride; or simply sending out information about Gaucher disease.

The IGA supports the event by providing downloadable materials and encourages its members to use social media

to share their stories and experiences. Patient Associations around the world will be supporting International Gaucher Day to:

- highlight the existence of Gaucher disease and its signs and symptoms to create greater awareness to ensure that patients are correctly diagnosed at an early stage to prevent delayed diagnosis
- inform communities of the existence of available treatments and clinical trials
- shine the spotlight on unmet medical need particularly the lack of treatment for patients with rare neuronopathic manifestations
- prioritise the tragedy of countless patients in many parts of the world who are unable to access life-saving treatments
- encourage further research into Gaucher disease with particular reference to the links that are emerging between Gaucher disease and more common conditions such as Parkinson's disease.

Posters and factsheets but are available to download from the website

https://gaucheralliance.org/gb/international_gaucher_day_rare_but_not_alone

EWGGD 2019 Scientific meeting



The 13th EWGGD Meeting was held in Clermont-Ferrand (France) from 3 to 6 July 2019. This conference is the only academic congress dedicated to Gaucher disease, bringing together clinicians and scientists, who are experts in Gaucher disease, and the international patient association, the IGA (International Gaucher Alliance). This congress traditionally takes place every two years.

This year the congress brought together 38 nationalities with talks given by 26 faculty and moderated by 16 experts. The Scientific Committee [Prof. S. Vom Dahl (Düsseldorf, Germany), Prof. M. Horowitz (Tel Aviv, Israel), Prof. H. Aerts (Amsterdam, Netherlands), Prof. M. Berger (Clermont-Ferrand, France): host] selected 26 oral communications from the 91 papers submitted.

The rich program focused on the main challenges related to this condition. Despite the therapeutic arsenal available to the Gaucher Community, many questions remain unanswered. Following the special opening lecture by Prof. T.M. Cox (Cambridge, UK), the program was composed of 8 sessions with the following topics: Phenotypic heterogeneity/ Neurological disorders/ Hematological-visceral form: Is everything known ?/ Biomarkers/ Bone disease, what's new?/ Access to care/ Personalization of treatment / Perspectives. Each session provided a scientific review of the subject, highlighting questions and important objectives

Studies carried out by teams around the world substantiate the existence of marked phenotypic heterogeneity, a synthesis of which was carried out by Prof. P. Mistry (Yale University, New Haven) and the role of genetic modifiers was discussed by Prof. E. Sidransky (Bethesda). With regard to neurological impairment, after a description of the neurological symptoms of patients with type 1 and type 3 Gaucher disease (Dr. M. Machaczka, Stockholm), the relationship of GD with Parkinson's disease was discussed by Prof. M. Horowitz with particular reference to the Drosophila model.

After an update study on the functioning of the spleen (Dr P. Amireault, Paris), abnormalities of the erythroblastic line were discussed (Dr M. Franco, Paris). Two exciting papers concerned the antigenic targets of monoclonal immunoglobulins in Gaucher (KD. Preuss, Homburg and S. Nair, New Haven); Many questions remain on this fascinating subject and we look forward to updates in the future.

The session on biomarkers (Prof. H. Aerts, Amsterdam

26 Invited Speakers

26 Oral Communications

63 Posters

235 Attendees

38 Nationalities.

and Prof. P. Deegan, Cambridge) put into perspective the practical application of biomarkers and asked explored the challenge of optimization of the use of current biomarkers in order to be able to further personalize patient

management. In the bone pathology session Prof. D. Hughes (London, UK) reviewed the impact of glucocerebrosidase deficiency on osteoblast and osteoclast metabolism, before Prof N. Belmatoug (Paris, France) synthesized medical and radiological monitoring of Gaucher bone disease.

The patient associations, lead a very informative discussion on access to care, reiterating the important heterogeneity of access to care and showing how much progress is to be made. Eugen Mengel (Germany) and Carla Hollak (Amsterdam, Netherlands) organized a stimulating debate on the issue of dosage and duration of treatment whilst Prof. E. Lukina, (Moscow) updated us on eligibility and Prof. R Shiffman on venglustat

We were also delighted to have presentations on new developments by young researchers addressing new ex vivo or animal models, and the identification of new aspects of enzyme deficiency on cell metabolism, allowing us to deepen our knowledge on the pathophysiology of this disease and new approaches to therapy such as gene therapy .

Finally, in the closing session, Prof. James Shayman (Michigan, USA) discussed strategies to develop new molecules capable of crossing the blood-brain barrier.

Oral and poster presentations demonstrated the dynamism of research on Gaucher disease and the emergence of young researchers. We would like to thank all the teams that submitted work, which considerably enriched the content of the congress and the discussions.

The majority of the speakers agreed to put their slideshows online (see the congress website by following the link: <https://presentations.ewggd2019.com/2019>).

During this congress, 4 prizes were awarded:

Three prizes were given for oral communication, one from the IGA and two from the EWGGD. The IGA award was



presented to Aimee Donald, Salford, UK. Two EWGGD awards were given to Eleonora Pavan, Udine, Italy and Daria Messelodi, Bologna, Italy. The EWGGD poster awards were given to Gizem Onal, Ankara, Turkey

and Daphne Boer, Leiden University, Netherlands. We warmly congratulate the winners!

In addition to this scientific program, a social program was organized to discover Clermont-Ferrand and the Auvergne region, and in particular the oldest volcanoes in Europe making up the Puys chain, classified as a UNESCO World Heritage Site since 2017, with two dinners in remarkable sites.

We would like to thank all the speakers, participants and organizers of this congress who made it possible to create a friendly, warm and stimulating atmosphere; it was a great pleasure to welcome them to Clermont-Ferrand.

Prof. Marc Berger, host, on behalf of 13th EWGGD Meeting Scientific Committee

Meet the board:

Our new board was elected at the 2019 business meeting. We are privileged to have the ongoing support of our founding president Prof Hans Aerts and our IGA chair Mr Jeremy Manuel. Prof Marc Berger remains on the board as vice chair and we welcome Prof Derralynn Hughes as Chair, Prof Shoshana Revel-Vilk as secretary, and Drs Andrea Dardis and Maciej Machaczka as board members.

EWGGD board 2019
Chair : Prof Derralynn Hughes UK
Vice Chair : Prof Marc Berger France
Founding president : Prof Hans Aerts Netherlands
IGA: Mr Jeremy Manuel UK
Dr Andrea Dardis Italy
Dr Shoshana Revel-Vilk Israel
Dr Maciej Machaczka Sweden

The board is keen to hear from you so please do get in touch! In future newsletters we will be interviewing our board members and learning about their interests and priorities in relation to Gaucher disease.

EWGGD New Activities:

We aim to make the EWGGD a real working group promoting and facilitating research, education and clinical support for the Gaucher community






New activities include the following:

- 1) A monthly newsletter to keep you upto date
- 2) A brand new EWGGD website coming soon!
- 3) Scientific working groups: you are invited to join one of our scientific working groups to work together to create

policy statements, guidelines and to facilitate research. We will be working throughout the year and hope to devote time at the next scientific meeting to the scientific working groups. Watch out for topics for the working groups in our next newsletters and please do suggest topics for consideration.

4) Monthly educational webinars for those new and established in the GD community. The content to be shared on our website.

5) Working with external organisations and collaborators to further the aims and benefit to the Gaucher community, including IGA, EHA, and industry. We are currently working on a position statement related to working with industry and will share this with you all shortly

EWGGD - The European Working Group on Gaucher Disease				
Connection and Communication				
Collaboration				
Patient Focus				
Connectivity and visibility	Collaboration	Education and training	Policy	Research
 <ul style="list-style-type: none"> Website News bulletin Monthly webex Generic email address Branding Biannual meeting Congress presence Publications 	 <ul style="list-style-type: none"> IGA Workshops Working groups EHA ERN Speciality-specific groups Industry 	 <ul style="list-style-type: none"> Education platform Slide bank Educational webex Support for young investigators Mentoring Preceptorships 	 <ul style="list-style-type: none"> What are the needs? Guidelines? Ethics? Data sharing consent Gene therapy Working with industry Working with regulators 	 <ul style="list-style-type: none"> What are the questions? Research platform Clinical trial network Biobanking Sample sharing Lab research Joint grant applications Outcome measures Qualitative research
EWGGD2019@gmail.com				

Dates for your Diary:

First Educational webinar: Introduction to Gaucher Disease: Clinical Manifestations: Prof Derralyann Hughes 1730 CET (1630 UK) Wednesday 6th November 2019
Link : <https://zoom.us/j/555501579>

Newsletter: We are hoping to provide you with a newsletter around 1st of each month. If you have news, announcements and contents to share please email ewggd2019@gmail.com by 18th day of the preceding month

2020 EWGGD Scientific meeting Amsterdam: watch out for the dates coming soon

Get involved:

This is your EWGGD and we would love the whole community to be involved. There is so much to be done and we need many hands to help!

Please let us know if you would like to be involved with any of the following:

- Share your news and meetings in the EWGGD newsletter
- Suggest a title for an educational webinar or volunteer to give one of our webinars
- Tell us about your research
- Volunteer for one of our scientific working groups

Logo Competition!



Please help us to refresh the EWGGD branding and logo. Please send your suggestions for illustrations and branding to help design our newsletter and website. Consider getting your students and young people involved! Closing date 30 November 2019. The board will consider entries at the December board meeting and winner(s) announced in January 2020

Membership:

We are in the process of contacting our existing members, supporters and meeting participants to confirm their membership, we hope the new website will provide a portal for membership. If you are not a member and would like to join then email ewggdmembership@gmail.com

Contact us:

Email: ewggd2019@gmail.com

Membership enquiries: ewggdmembership@gmail.com

Follow us on Twitter @EWGGD1