12-13 OCTOBER 2020

European Working Group on Gaucher Disease

14th EWGGD MEETING

SCIENTIFIC PROGRAMME

Host: Professor Hans Aerts,
Leiden Institute of Chemistry,
Leiden University,
The Netherlands

www.EWGGD2020.com

12-13 OCTOBER 2020

European Working Group on Gaucher Disease

14th EWGGD MEETING

CONTENT

WELCOME MESSAGE

Dear Friends, Colleagues, Patients,

On behalf of the EWGGD board, we are happy to welcome you to the 14th European Working Group on Gaucher Disease meeting, which is taking place exceptionally this year in a virtual format.

While we are saddened we cannot meet you in person and spend the next 2 days together, we are also happy to have this opportunity to celebrate together the 25th year of the EWGGD with this virtual meeting, and we trust that the scientific content and the discussions will still be of the highest quality.

We thank everyone taking part in this meeting over the next 2 days; all the international speakers, in particular those who are connecting in the very early hours in the morning due to the time zone differences, the IGA, patient advocates, poster presenters and industry partners for your participation and support.

We look forward to the next few days and we invite you to engage with our faculty, patients and partners while participating actively in the programme sessions.

Thank you for joining us this year as we continue to explore the vast world of Gaucher Disease,

With our best wishes,

Derralyn Hughes
EWGGD Chairperson

Hans Aerts
EWGGD Board Member and
Host of EWGGD 2020

SCIENTIFIC COMMITTEE

Professor Hans Aerts

Department of Medical Biochemistry,

Leiden Institute of Chemistry, Leiden University (*The Netherlands*)

Professor Marc Berger

Department of Hematology (*Biology*)

CHU Estaing, Clermont Auvergne University (*France*)

Doctor Andrea Dardis

Centre for Rare Disease, Genetics

Azienda Sanitaria Universitaria Integrata di Udine (*Italy*)

Professor Derralynn Hughes

Department of Hematology

Royal Free Hospital (*United Kingdom*)

Professor Shoshana Revel-Vilk

Gaucher Unit, Pediatric Hematology

Shaare Zedek Medical Center (*Israël*)

European Working Group on Gaucher Disease

14th EWGGD MEETING

Host: Professor Hans Aerts, Leiden Institute of Chemistry, Leiden University (The Netherlands)

CELEBRATING 25 YEARS OF THE EWGGD!

October 12-13 2020

E-CONGRESS

Scientific Committee: **Johannes Aerts,** *The Netherlands;* **Marc Berger,** *France;* **Andrea Dardis**, *Italy;* **Derralynn Hughes,** *United Kingdom;* **Shoshana Revel-Vilk,** *Israël*

FINAL PROGRAMME

Please note that the programme is scheduled according to Central European Time (CET)

Monday, October 12, 2020

09:00 – 12:00	EWGGD WORKING GROUP SESSIONS *Pre-registration required*
09:00 – 10:30	**Laboratory**
10:30 – 11:30	**Clinical Working Group Discussions** Room 1: Room 1: Diagnosis Room 2: Treatment and monitoring Room 3: Diagnosis and management of comorbidities/disease related complications/life events Room 4: Outcome that are important to patients and universal issues related to GD.
11:30 – 12:00	**Clinical Working Group Summary**

12:00 – 13:00 | BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL

13:00 – 14:00	OPENING SESSION	***Chairpersons:*** Hans Aerts, (*Leiden NL*), Derralynn Hughes *(London, UK)*
13:00 – 13:25	Opening speech	Hans Aerts, (*Leiden NL*), Derralynn Hughes *(London, UK)*
13:25 – 13:55	Keynote lecture: GBA deficiency: Gaucher disease	Ari Zimran *(Jerusalem, IL)*
13:55 – 14:00	Live Q&A	Ari Zimran *(Jerusalem, IL)*

14:00 – 14:15 | BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL

14:15 – 15:30	SESSION 1 : NOVEL OR INVESTIGATIONAL THERAPIES	***Chairperson:*** Derralynn Hughes *(London, UK)*
14:15 – 14:30	Introduction	Derralynn Hughes *(London, UK)*
14:30 – 14:40	54: In utero enzyme replacement therapy in fetuses with neuronopathic Gaucher disease: a phase I clinical trial	Marisa Schwab *(San Francisco, US)*
14:40 – 14:50	4: PR001 gene therapy improved neuronopathic Gaucher disease phenotypes in mouse models by increasing GCase activity	Patty Sheehan *(New York, US)*
14:50 – 15:00	19: The Guardone clinical trial protocol: a first-in-human, open-label, multinational phase 1/2 study of avr-rd-02 ex vivo lentiviral vector, autologous gene therapy for gaucher disease	Leslie Jacobsen *(Cambridge, US)*
15:00 – 15:10	38: Venglustat combined with Imiglucerase positively affects neurological features and brain connectivity in adults with gaucher disease type 3	Timothy Cox *(Cambridge, UK)*
15:10 – 15:18	20: Outcomes after 4.5 Years of Eliglustat in Treatment-naïve Adults with Gaucher Disease Type 1 in the Phase 3 ENGAGE Trial: Aggregate and Individual Patient Responses	Pramod Mistry *(New Haven, US)*
15:18 – 15:30	Live Q&A	All

15:30 – 16:00 | BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL

16:00 – 17:00	**SESSION 2 : CLINIC**	***Chairpersons:*** Shoshana Revel-Vilk *(Jerusalem, IL)* & Maciej Machaczka *(Stockholm, SE)*
16:00 – 16:10	53: Liver steatosis is associated with metabolic risk factors and liver fibrosis in adult patients with type 1 gaucher disease	Francesca Carubbi *(Modena, IT)*
16:10 – 16:20	12: Hematologic malignancies and gammopathies in gaucher disease type 1	Maria Cappellini *(Milan, IT)*
16:20 – 16:30	15: Delineating D409H (D448h) homozygous phenotype-genotype in an international cohort of the ICGG gaucher registry: cardiac involvement and early mortality	Hagit Baris-Feldman *(Tel Aviv, IL)*
16:30 – 16:40	30 & 31: Cognitive decline and depressive symptoms; early non-motor presentations of Parkinsonism among Egyptian Gaucher patients & Retinal thinning in Adolescents and Young Adults with Gaucher Disease; Relation to Parkinsonian Features	Amira Adly *(Cairo, EG)*
16:40 – 16:50	44: Long term follow up of osteopenia in Egyptian type 3 Gaucher disease and its response to Enzyme Replacement Therapy (ERT) and bisphosphonates	Magy Abdelwahab *(Cairo, EG)*
16:50 – 17:00	Live Q&A	All

17:00 – 17:30	**YOUNG RESEARCHERS**	***Chairpersons:*** Marc Berger (*Clermont-Ferrand, FR)* & Andrea Dardis (*Udine, IT)*
Introduction to the network for young researchers		Majdoleen Istaiti (*Jerusalem, IL*), Lucia Lavalle (*London, UK*); Marta Artola (*Leiden, NL,*); Dr. Eleonora Pavan (*Udine, IT*)

17:30 – 18:10	**KEYNOTE LECTURE**	
17:30 – 18:00	Understanding GBA1-associated Parkinsonism: Advances and challenges	Ellen Sidransky *(Washington, US)*
18:00 – 18:10	Live Q&A	Ellen Sidransky *(Washington, US)*

18:10 – 19:00 | VIRTUAL EXHIBITION AND ABSTRACT HALL

ADJOURN

Tuesday, October 13, 2020

08:00 – 08:50 | VIRTUAL EXHIBITION AND ABSTRACT HALL

8:50 – 9:00	DAY 2 INTRODUCTIONS	Hans Aerts *(Leiden, NL)*

9:00 – 10:00	SESSION 3 : LABORATORY	***Chairpersons:*** Marc Berger (*Clermont-Ferrand, FR)* & Andrea Dardis (*Udine, IT)*
09:00 – 09:10	17: Gba1 inactivation in oligodendrocytes affects myelination and induces hallmarks of neurodegeneration in mice	Matilde Cescon *(Padova, IT)*
09:10 – 09:20	42: Integrated approach of clinical exome sequencing and MLPA assay for the molecular analysis of GBA gene.	Stefania Zampieri *(Udine, IT)*
09:20 – 09:30	46: Beta-Xylosidase and transxylosidase reactions of human glucocerebrosidase	Hans Aerts (*Leiden, NL)*
09:30 – 09:40	14: Prospective study of the natural history of chronic acid sphingomyelinase deficiency in children and adults during 11 years of observation	Margaret Mcgovern *(New York, US)*
09:40 – 10:00	Live Q&A	

10:00 – 10:30 BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL

10:30 – 11:35	IGA SESSION	
10:30 – 10:40	Regional Manager programme	*Suyog Sathe (IGA*)
10:40 – 10:55	Raising awareness and due diligence in Nepal	Ashok Vellodi (Bushey, UK)
10:55 – 11:10	Developing PROs for nGD	Elin Haf Davies (London, UK)
11:10 – 11:20	GD1 Guidelines initiative	Chris Hendriksz (Manchester, UK)
11:20 – 11:25	Comorbidities in Gaucher disease, PD/GD	Margaret Giuliani *(IGA)*
11:25 – 11:30	Older Generation	Biljana Jovanović *(IGA)*
11:30 – 11:35	Live Q&A	

11:35 – 12:05	KEYNOTE LECTURE	
11:35 – 11:55	GBA deficiency: Malignancies	Carla Hollak *(Amsterdam, NL)*
11:55 – 12:05	Live Q&A	Carla Hollak *(Amsterdam, NL)*

12:05 – 13:00 BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL

13:00 – 14:30	**SESSION 4 : POSTER HIGHLIGHTS** ***Chairpersons:*** Shoshana Revel-Vilk *(Jerusalem, IL)* & Marc Berger (*Clermont-Ferrand, FR)*	
13:00 – 13:05	2: Alpha-Synuclein dimerization in erythrocytes of gaucher disease carriers and patients before and after enzyme replacement therapy	Marina Moraitou *(Athens, GR)*
13:05 – 13:10	7: Parkinsonism in type 3 Gaucher disease: Three cases highlighting the clinical phenotype and presentation of Parkinson disease in a rare cohort.	Emory Ryan *(Betheshda, US)*
13:10 – 13:15	11: Identification of risk features for complication in Gaucher's Disease patients: A Machine Learning analysis of the Spanish Registry of Gaucher Disease	Marcio Andrade-Campos *(Feeteg, ES)*
13:15 – 13:20	16: Autophagic impairment and differentiation delay in an in vitro model of Schwann cells lacking GBA1	Loris Russo *(Padova, IT)*
13:20 – 13:25	21: Serum progranulin levels in paediatric patients with gaucher disease; relation to disease severity and liver stiffness by transient elastography	Azza Tantawy (Cairo, EG)
13:25 – 13:30	24: Next generation sequencing for newborn lysosomal storage screening	Laura López De Frutos *(Zaragoza, ES)*
13:30 – 13:45	Live Q&A	All
13:45 – 13:50	28: The impact of covid-19 pandemic on Brazilian patients with gaucher disease	Ida Schwartz *(Porto Alegre, BR)*
13:50 – 13:55	33: Outcomes of pregnancies in patients with Gaucher Disease: The experience of a center of excellence on rare metabolic disease-Gaucher disease, in Greece	Komninaka Veroniki *(Athens, GR)*
13:55 – 14:00	36: Surrogate markers of bone disease response to treatment. Data from a gaucher disease reference centre in Spain	David Moreno-Martinez *(Barcelona, ES)*
14:00 – 14:05	41: Development of brain penetrant non-competitive pharmacological chaperones for the treatment of neuronopathic gaucher disease	Natalia Perez *(Barcelona, ES)*
14:05 – 14:10	48: Switching between ERT and SRT in patients with gaucher disease: data from the gaucher outcome survey (GOS)	Derralynn Hughes (London, UK)
14:10 – 14:15	49: Multi-parameter, neurological, prospective study (SENOPRO_GAUCHER study) of type 1 Gaucher Disease patients: preliminary results.	Emanuele Cerulli Irelli *(Rome, IT)*
14:15 – 14:30	Live Q&A	All
14:30 – 15:00	**BREAK, VIRTUAL EXHIBITION AND ABSTRACT HALL**	
15:00 – 15:20	**ABSTRACT COMPETITION ANNOUNCEMENTS** ***Chairpersons:*** Hans Aerts, (*Leiden NL)*, Derralynn Hughes *(London, UK)*	
15:20 – 16:00	**KEYNOTE LECTURE**	
15:20 – 15:50	Bone metabolism and pathophysiology: *Lessons from the Diagnosis and Treatment of Metabolic Bone Disease*	Richard Eastell *(Sheffield, UK)*
15:50 – 16:00	Live Q&A	Richard Eastell *(Sheffield, UK)*
16:00 – 16:15	**CLOSING REMARKS FROM EWGGD 2020 HOST & EWGGD CHAIRPERSON** ***Chairpersons:*** Hans Aerts, (*Leiden NL)*, Derralynn Hughes *(London, UK)*	
	ADJOURN	

The EWGGD 2020 meeting endeavours to put the patient at the centre of discussions from the early stages of the planning to the end.

The EWGGD 2020 certify that the 2020 EWGGD conference meet the five Patients Included Conference Charter Clauses:

1. Patients or caregivers with experience relevant to the conference's central theme actively participate in the design and planning of the event, including the selection of themes, topics and speakers.

- Patients and patient representatives are very present among the speakers and audience

2. Patients or caregivers with experience of the issues addressed by the event participate in its delivery, and appear in its physical audience.

- Patients and patient representatives and caregivers are among EWGGD 2020 faculty members and form a large part of the audience

3. Travel and accommodation expenses for patients or carers participating in the advertised programme are paid in full, in advance. Scholarships are provided by the conference organisers to allow patients or carers affected by the relevant issues to attend as delegates.

- Travel and accommodation is assured for patients, patient representatives and caregivers who form part of the faculty
- Grants are made available for patients, patient representatives and caregivers upon application to attend as delegates

4. The disability requirements of participants are accommodated. All applicable sessions, breakouts, ancillary meetings, and other programme elements are open to patient delegates.

- A dedicated assistive personnel accompanies the patients if required to ensure their comfort and that their needs are accommodated throughout the conference.
- A room in the conference centre is available for patients to rest during the day
- The transport is secured from and to the hotel
- The venue was chosen to have high accessibility

5. Access for virtual participants is facilitated, with free streaming video provided online wherever possible.

- The presentations will be published online on our website and are accessible to all

THANK YOU TO OUR SUPPORTERS

PLATINUM

SANOFI GENZYME

Takeda

GOLD

Pfizer

Breakthroughs that change patients' lives

INTERNATIONAL GAUCHER ALLIANCE

IGA INTERNATIONAL GAUCHER ALLIANCE
Driven by passion for patients

European Working Group on Gaucher Disease

14th EWGGD MEETING

Host: Professor Hans Aerts, Leiden Institute of Chemistry, Leiden University (The Netherlands)

GENERAL INFORMATION

EWGGD The European Working Group on Gaucher Disease

The EWGGD represents clinicians and researchers with an interest in Gaucher disease. In February, 2012 the Group reached a formal status. The EWGGD is affiliated to the International Gaucher Alliance (IGA), formally the European Gaucher Alliance (EGA) representing the interests of the patients. This alliance was formed shortly after the founding of the EWGGD and reached a more formal status in 2008, when during the EGA meeting patient representatives of 24 countries unanimously agreed to incorporate the EGA into a formal charitable body, to speak for Gaucher patients on a Pan European level.

For more information about becoming a member please visit the website

www.ewggd.com

IGA INTERNATIONAL GAUCHER ALLIANCE
Driven by passion for patients

About International Gaucher Alliance (IGA)

Why do we need an international umbrella group? The working together of Gaucher patient groups has already lead to consistency in treatment of certain manifestations of Gaucher Disease throughout Europe. Inspired by this and by their successes in achieving humanitarian aid for the most severe patients in Eastern Europe member countries of the IGA has grasped the challenge of collaboration for the further advantage of Gaucher Patients.

Although the most common Lysosomal Storage Disorder Gaucher Disease is very rare with an incidence of about 1 in 100,000 live births.

Patient Groups have worked closely together to share their experiences and to facilitate access to treatment. Scientists and Clinicians throughout the world increasingly collaborate both in research and in the development of strategies for the management of the disease.

Pharmaceutical companies are Global and clinical trials are frequently multi-centred with treatments being approved in different countries worldwide, while in others patients are not able to benefit from them.

Patient Groups throughout the world cannot work in isolation and their alliance by joining the IGA provides a clear global voice of representation and opportunity for advancement.

www.gaucheralliance.org

LANGUAGE

English is the official language of the congress

ACCREDITATION

The 14th Meeting of European Working Group on Gaucher Disease 12/10/2020-13/10/2020 has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with **10** European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

Through an agreement between the Union Européenne des Médecins Spécialistes and the American Medical Association, physicians may convert EACCME® credits to an equivalent number of AMA PRA Category 1 Credits ™. Information on the process to convert EACCME® credit to AMA credit can be found at www.ama-assn.org/education/earn-credit-participation-international-activities.

Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME® for ECMEC®s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

CME certificates will be emailed to all participants after the meeting after completion of the evaluation form.