In 2019 the International Gaucher Alliance continued to grow in membership as we welcomed: Australia & New Zealand, Brazil, Tunisia, Colombia and new patient groups from the US and Croatia, taking our membership up to 55 patient associations from 54 countries: we are now advocating for Gaucher patients in 5 continents!

Our growth is also seen as we expand our activities in parts of the world where there are no organizations to provide support to many patients and families. We are extremely proud of our educational activity in Nepal where with our Regional Managers program we managed to educate over 100 doctors from 3 different cities; Kathmandu, Dharan and Biratnagar on Gaucher and other Lysosomal storage disorders.

In the research area, we continued to take forward the development of a global patient led nGD registry and we are excited that we will work in partnership with Kantar on this project who will run the day to day logistics of the registry, which will be an important project for nGD patients and families.

The IGA will continue to be innovative, patient-centered and focused on supporting organizations for Gaucher disease all over the globe. We grow together with our members. We are stronger when the patients we support become stronger. The future will bring a change for the better.

VESNA ALEKSOVSKA (CHAIR)     TANYA COLLIN-HISTED (CEO)

Let’s continue to work together as we are rare but not alone.
IN 2019 WE MADE THE DIFFERENCE BY:

- Helping to provide treatment to 37 patients following requests from 52 patients in 11 countries
- Raising awareness about Gaucher disease through International Gaucher Day – on 1 October we were able to reach over 6000 people on Twitter; over 8000 people on Instagram and over 12,500 people on Facebook
- Facilitating and participating in a three-day educational programme in Morocco that attracted 41 delegates from Ghana, Morocco, Tunisia, Kenya, Rwanda, Niger, Ivory Coast, Congo and Mauritania
- Organizing our first ever educational event on Lysosomal Storage Disorders in 3 cities of Nepal – Kathmandu, Dharan and Biratnagar: attended in total by over 100 doctors
- Starting the development of a global disease registry for neuronopathic Gaucher disease and accomplishing phase I of the registry in partnership with parent/patients and carers; key opinion leaders and four industry partners
- Presenting at more than 6 various international symposiums and conferences on different subjects reaching an audience of over 900 doctors, researchers, nurses, patient advocates and patients
- Collaborating with 8 pharmaceutical companies regarding the development of potential new treatment options for Gaucher patients
- Attending patient meeting in Albania and supporting the development of a new patient group in this country
- Appointing 2 new regional managers to increase our footprint in Central America and Caucasus & Central Asia with a commitment to Africa for 2020

CHARITABLE ACCESS PROGRAMME

We have always pledged to help any Gaucher patient that asks us for help wherever they live in the world, therefore advocating for charitable treatment has always been and will remain our priority.

Despite there being three licensed enzyme replacement therapies (ERT) and two substrate reduction therapies (SRT) available for the treatment of the visceral manifestations of the disease, sadly, there are still hundreds of patients around the world without access to treatment.

In 2019, we received 52 direct requests for treatment from 11 countries such as Tunisia, Tanzania, Pakistan, Sudan, Kenya, Algeria, Qatar, Rwanda, Iran, India, Morocco. By working closely with Sanofi Genzyme and Takeda we helped to give a future to 37 Gaucher patients as they received donated ERT.

In 2019, we made the difference by:

We supported a one-day meeting/clinic in Jordan for patients with Type 3 Gaucher disease and their families in collaboration with the Jordanian Gaucher association to discuss Pfizer GD 3 clinical trial with Elelyso.

Unfortunately, there are many more other patients suffering without treatment.

Currently, we are aware of at least 250-300 patients, mainly children, waiting to get treatment through charitable programmes.
RAISING AWARENESS:
INTERNATIONAL GAUCHER DAY (IGD)

International Gaucher Day, first launched by the IGA in 2014, is celebrated annually on 1st October. This year we asked members of the Gaucher community to "mark it with ink!" and wear temporary IGD tattoos in order to spread the word about the disease and to raise awareness.

In 2019 our theme was 'Rare Stars - Bringing hope, improving lives' with a personal focus on those unseen rare stars who had shown support to Gaucher patients during their journey. All nominations received were included in a montage published on 1 October.

During the EWGGD meeting in Clermont-Ferrand (France), we unveiled our own Rare Star: Jeremy Manuel, OBE in recognition of his 25 years' dedication to the global Gaucher community.

Throughout the 50+ member countries of the IGA patients, families, doctors, scientists and healthcare professionals marked IGD by holding events and activities using downloadable promotional materials made available through the IGA office.

We were absolutely delighted with all the engagement on social media for International Gaucher Day.

EDUCATING DOCTORS

These words expressed by Hassan Suraw, a father of a little girl suffering from Gaucher in Kenya, describe our strong motivation and determination to support access to education and training for doctors, in particular young doctors in countries where there is limited knowledge and where doctors only have one or a few Gaucher patients.

In March we took part in Gaucher and Fabry preceptorship at the Royal Free Hospital in London.

In May we participated in FYMCA's meeting in Morocco attended by 41 doctors from 9 different countries, from all over West Africa.

MEDICAL PROGRAMME IN NEPAL

In September we organized a three-day educational programme on Lysosomal Storage Diseases (LSDs) for doctors in Nepal. Unfortunately, the awareness of LSDs among the doctors is extremely low - the fact that no Gaucher patient has been diagnosed in this country speaks as proof of that. The IGA was represented by our two regional managers Shashank Tyagi and Suyog Sathe and educational training was carried out by Dr. Ashok Vellodi who gave lectures covering case-based approaches, accurate diagnosing and better management of LSDs.

Educational training was carried out in three cities: Kathmandu (attended by over 53 doctors), Dharan (attended by 24 doctors) and Biratnagar (attended by 27 doctors).

"She had to go through so much pain because the doctors didn't know what was wrong with her" - The slogan of IGD is RARE BUT NOT ALONE

This activity not only raised awareness and identified doctors willing to develop an interest in LSDs, but also revealed numerous challenges and various problems to be tackled in order to help patients. Most significant of them are:

• lack of coordination, knowledge-sharing and common practices among the doctors from different cities in Nepal;
• lack of genetic training and awareness among doctors;
• lack of awareness and knowledge among parents/caregivers;
• need for a patient support group;
• lack of diagnostic facilities and infrastructure in Nepal;
• need for charitable access programmes for diagnosed patients.

We would like to thank Nepal Paediatric Society, Maharajgunj Medicine Campus and Tribhuvan University Teaching Hospital for their support with arranging these events.

All participating doctors recognized that this educational activity improved their knowledge and understanding of symptoms, clinical diagnosis and management of LSDs. Furthermore, they would be willing to work together with us.

The staff from B.P. Koirala Institute of Health Sciences in Dharan, Nepal.
Our regional managers are our “eyes and ears” in the regions of the globe where there is limited/poor patient activity. They are proactive and interact with the key stakeholders in these Gaucher communities and serve as the main contact point for patients, the pharmaceutical industry and the healthcare professionals in the region, where no member organisation is present.

In 2019 our first regional managers—Marketa, Shashank and Suyog—were present. In 2020, our first regional manager in Latin America was Patricia with the support of Adel. Our regional managers work hard to improve patient care and to establish dialogue between doctors, patients & the government.

"During my first month working as an IGA regional manager, I had the opportunity of getting acquainted with other organisations around the world and have already attended a meeting in Geneva with Gaucher specialist which enabled me to bring back new knowledge that I will pass on to doctors in Eastern Europe and Mongolia, and I hope, this will open opportunities to share experience with other professionals in this region, especially with the doctors in Dominican Republic. I am excited about the projects for 2020 and I believe that the work of regional managers will enrich the knowledge and bring different perspectives to an already positive work developed over the years."

—Marketa, RM in Caucasus & Central Asia

PATIENT INVOLVEMENT:

10 volunteers via focus group and in-depth interviews
On-line Delphi with 23 respondents from 13 countries

1 MEETING 5 KOLs from 3 countries

"It is truly wonderful to be part of this patient driven initiative. Engaging innovative approaches and global collaborative efforts in this way, with early dialogue among all the players will be sure to help move the needle significantly in this space.”

—Dr Elin Haf Davies, CEO of aparito

GLOBAL DISEASE REGISTRY FOR NEURONOPATHIC GD:

Neuropathic Gaucher Disease (nGD) has a high unmet need, but with an increasing number of pharmaceutical companies now developing treatment options, it brings hope to many patients & families.

A collaborative disease registry will:

• offer important insight for understanding the natural history of the disease
• correlate global phenotypes and genotypes
• validate new endpoints/assessments
• support clinical trial designs
• generate a data source that can be used for both Regulatory and Health Technology Assessments evaluation of emerging drugs for nGD.

The IGA acted as the lead instigator, bringing together all the partners with funding from the pharmaceutical industry and support from aparito to explore patient, KOL, and pharma needs and requirements.

It took 16 weeks to accomplish phase 1 of the development of this global patient registry, with 132 clinical data fields identified to be included in the registry.

PATIENT INVOLVEMENT:

• 10 volunteers via focus group and in-depth interviews
• On-line Delphi with 23 respondents from 13 countries

We worked with aparito and Phase I was supported with unrestricted grants from four pharmaceutical companies: Oxyrane, Pfizer, Prevail and Sanofi Genzyme.

"It is truly wonderful to be part of this patient driven initiative. Engaging innovative approaches and global collaborative efforts in this way, with early dialogue among all the players will be sure to help move the needle significantly in this space.”

—Dr Elin Haf Davies, CEO of aparito
**INFLUENCING GAUCHER RESEARCH AGENDA:**

As we continue to serve the Gaucher community, it is our priority to work to ensure that the Gaucher research agenda is focused on addressing key unmet needs from a patient perspective.

To achieve this aim, we are actively involved at various international symposiums devoted to Gaucher or rare diseases in general. Our aim always is to show the perspective of the patients and patient advocates interested in Gaucher disease and to ensure that the patient is truly at the centre of everything they do.

Our poster award is given to a young health care professional during the EWGGD conference. We have chosen to give this award to encourage young healthcare professionals to become more involved and engaged in the field of Gaucher disease.

We set the specific criteria from year to year, however, significant benefit to patient care and/or health outcome in Gaucher disease should be demonstrated. In 2019 the criteria was “furthering the understanding of neuronopathic Gaucher disease and therefore improving patients’ lives”.

The winner of our award was Dr. Aimee Donald from Manchester University Hospital, UK.

The work presented was on the role of saccades in defining Gaucher Phenotypes. The award includes a diploma and a small grant.

Dr. Aimee Donald

"Receiving the IGA award for contributions to research in nGD was very unexpected. I still feel like I know so little about the disease but continue to learn every day. The welcome I’ve had within the community has been my motivation – especially the patients who share their stories with me and thank us so much. The award has enabled me to pursue another area of interest which I hope will benefit future work in nGD”

**FINANCE:**

The work of the IGA has been funded mainly by support from a number of pharmaceutical companies. We have also generated income through our CEO undertaking consultancy work and by grants from EURORDIS.

To ensure its independence, the IGA will only accept funds up to an amount not exceeding 35% of its annual budget from any one pharmaceutical company.

As a responsible organisation the IGA has a reserves policy to enable us, in the event of receiving no further funding, to continue to serve our global community and implement a strategy to address how to take the IGA forward.

**IT IS EQUAL TO £72,500**

We are extremely grateful for this dedicated support.

We thank very well all the people who finance this association because their good heart allows many children to survive around the world. It would not be possible to pursue this mission without your generous support.

**Thank you to our supporters…**

...for believing in us and sponsoring our extensive work program activities, various projects and events throughout the year. Without your generous support we couldn’t make a better world for Gaucher patients!

Amicus Therapeutics, Arbobio, Evotec, GS Abies, Orphazyme; Pfizer, Sanofi Genzyme, Takeda.

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**Grant & Corporate Funding**

33%

**Operational Costs**

17%

**Project Costs**

9%

**Volunteer**

20%

**Contributions: Institutions**

14%

**Consultancy**

71%
INSIGHT INTO WORKING AREAS FOR 2020/2021:

2020 will see the IGA:

Implement phase II of the global registry project

- Create a legal, governance, compliance and reporting structure that assures trust and quality for all.
- Seek regulatory buy-in (EMA/FDA) to the planned registry (technical and compliance).
- Ensure the strategic and financial support of all pharmaceutical companies that are developing treatments for nGD to make this a sustainable initiative.
- Create a technical platform today that can expand to meet the needs of the future, promoting FAIR data principles, and putting the patient at the centre.

Convene our biennial members meeting.

Launch our new website which will be more interactive and focussed on the needs of our visitors which are: the types of Gaucher disease and treatments available; getting access to treatment; finding a patient association; clinical research.

Organise the 2nd multi-stakeholder meeting with the main aim to publish a paper on international consensus guidelines on the clinical management of Type 1 Gaucher patients.

Provide educational opportunities to our membership through ‘town hall’ meetings.

Implement a structure to support Gaucher patients and families who are dealing with comorbidities and our first focus will be on Parkinson’s disease. The project aims to provide support and information on how best to manage these two chronic conditions and to develop a community for patients and their families with GD/PD and to identify the support needed.

Address the needs of the older generation of Gaucher patients (55+ years) by identifying and mapping the unmet needs, both medical and non-medical (i.e. social benefits), through questioning both the patients and physicians treating them. Based on the results, the IGA will address the challenges e.g. through the training of patients, member organisations and professionals.

Continue to raise awareness through our annual International Gaucher Day campaign.

2019 saw the IGA initiate in collaboration with Prof Huma Cheema from the Children’s hospital in Lahore, Pakistan and Mr Atif Qureshi from the Patient support group bring together representatives from the major stakeholders in the Gaucher community; Sanofi Genzyme, Pfizer, and Takeda to have an initial high-level meeting about their current activities in Pakistan, the challenges they face and their strategy going forward in an open and transparent way.

Identify ways to work together to support Prof Huma Cheema and her team in Pakistan and the Gaucher/LSD community. This was an agreed outcome from the IGA's 2019 initiative in collaboration with Prof Huma Cheema from the Children’s hospital in Lahore, Pakistan and Mr Atif Qureshi from the patient support group which brought together representatives from the major stakeholders in the Gaucher community: Sanofi Genzyme, Pfizer, and Takeda to have an initial high-level meeting about their current activities in Pakistan, the challenges they face and their strategy going forward in an open and transparent way. An outcome of this meeting was to have a face to face meeting in Lahore at the end of January 2020 with a few key opinion leaders, to meet with the Government and to develop a plan for Pakistan to seek ways collaboratively to improve access to medicines (all-encompassing diagnostics, education, treatment) for the Gaucher/LSD patients.