IGA INTERNATIONAL GAUCHER ALLIANCE

Driven by passion for patients

IMPACT REPORT 2019

Together we are stronger

INTRODUCTION:

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.

Yasmeen from Sudan

THE IGA:

The IGA is a patient led international organisation that has become the 'go to' global voice for over 85% of the Gaucher community and has built its reputation through listening to and delivering outcomes that have impacted on patients and their carers' lives.

OUR VISION:

A world where all Gaucher patients have access to the treatment and care they need and there is a possibility of a cure.

OUR MISSION:

- to **empower** its members
- to **advocate** on behalf of Gaucher patients
- to **ensure** that the Gaucher research agenda is **focused on patients' unmet needs**
- to **take collective action** to address challenges Gaucher patients worldwide face in accessing early diagnosis and optimal treatment and care.

OUR VALUES:

We are:

- **patient focused**
- **trustworthy**
- **collaborative**
- **visionary**
- **passionate**

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

In 2017 I found out that I have Gaucher disease. Because there is no treatment for Gaucher in Mongolia, me and my family found ourselves in a very difficult situation. Luckily, my sister learned about IGA and approached them asking for help. Thanks to IGA, I have been able to obtain the much needed treatment and my health is getting better. I would like to thank IGA for all the support I received and would like to wish the people who work there all the best!

Ariunbold Enkhbold, Mongolia

Morocco meeting

KEY ACTIVITIES:

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. Sadly, **8** patients who reached out to us for support died without getting access to the treatment they needed.

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. The purpose of the meeting was to inform the patients/parents and undertake genetic testing as confirmation of GD 3 and eligibility for the trial in partnership with the Shaare Zedek clinic in Israel.

Sadly, 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.

Nazlin (Kenya) pictured with her dad and brother

Sayyed (India)

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.

The slogan of IGD is
RARE BUT NOT ALONE

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

Greece IGD

EDUCATING DOCTORS

"She had to go through so much pain because the doctors didn't know what was wrong with her"

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).

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.

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, Maharajagunj Medicine Campus and Tribhuvan University Teaching Hospital for their support with arranging these events.

The staff from B.P. Koirala Institute of Health Sciences in Dharan, Nepal

EXPANDING OUR GLOBAL REACH TO LATIN AMERICA, CAUCASUS & CENTRAL ASIA:

Our regional managers are our "eyes and ears" in the regions of the globe where there is limited/no 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 joined by Patricia and Adel. Patricia supports patients in Central America; and Adel is responsible for Caucasus and Central Asia.

Our five regional managers are all Gaucher patients who perform this challenging role on a voluntarily basis. Special thanks to all of them for their work to make a difference to Gaucher patients in over **25** countries across the world.

"...the position of a regional manager is an opportunity to convey support to those who need it; those who have just encountered Gaucher disease or who live in a country where no one knows about this disease. I hope that I can build dialogue and collaboration between doctors, patients & the government."
(Adel Kaplan, our RM in Caucasus & Central Asia)

"During my first months working as an IGA regional manager I had the opportunity of getting acquainted with other organizations around the world and have already attended a meeting in Buenos Aires with Gaucher specialists which enabled me to bring back new knowledge that I could pass on to doctors in Guatemala and Nicaragua, and I hope, this will open opportunities to share experiences with other professionals in the 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." (Patricia Lucki, our RM in Central America)

GLOBAL DISEASE REGISTRY FOR NEURONOPATHIC GD:

Neuronopathic 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/outcomes
- 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 pharmaceutical needs and requirements.

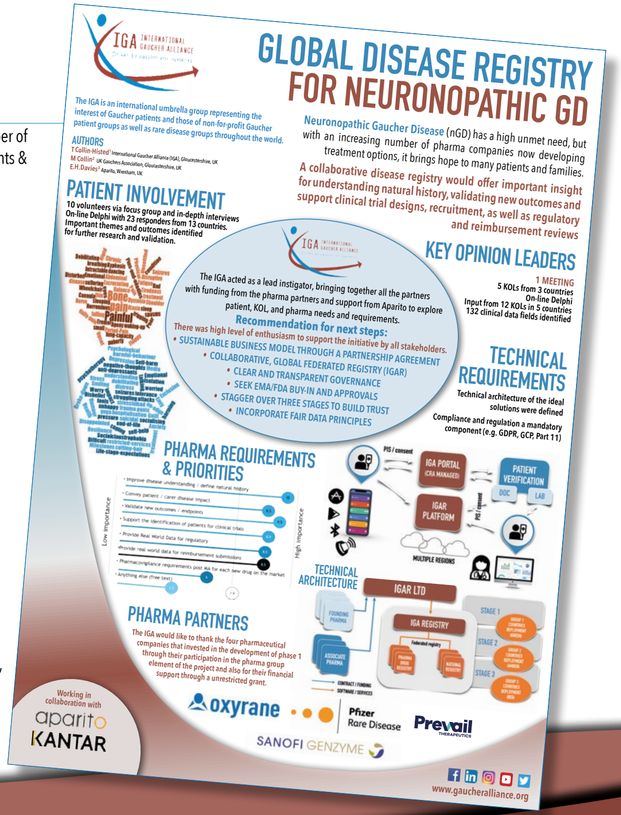
It took 16 weeks to accomplish phase I of the development of this global patient registry, with **132** clinical data fields identified to be included in the registry.

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 Therapeutics and Sanofi Genzyme.

"It is truly wonderful to be part of this patient driven initiative. Combining 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 and to address the challenges faced by them. The main conferences where we 'spread the word' were:

- 2nd International Congress on Advanced Treatments in Rare Diseases (RARE2019) speaking about the role of patient advocacy
- Takeda's 9th Gaucher Expert Summit, representing the global Gaucher patient voice
- Rare Disease International meeting, talking about the IGA's experience in patient-focused drug development
- the 13th European Working Group on Gaucher Disease (EWGGD), presenting on the IGA's role in helping the Gaucher community for accessing care, specifically via charitable access programme
- Cambridge Rare Disease Network (CRDN) RARESummit19, taking part in the panel discussion "Patient groups partnering in the drug development process"
- Sanofi Genzyme's 5th International Gaucher conference on bone disease running a workshop alongside Dr Patrick Deegan, focusing on the patient perspective on bone health in Gaucher disease
- Sanofi Genzyme Patient Registry Council meeting

Attending these types of meeting is extremely important for the IGA as we are working hard to position ourselves as the 'go to' organisation for all stakeholders interested in Gaucher disease and to ensure that the patient is truly at the centre of everything they do.

IGA AWARD:

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.

"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 teach me so much. The award has enabled me to pursue another area of interest which I hope will benefit future work in nGD"

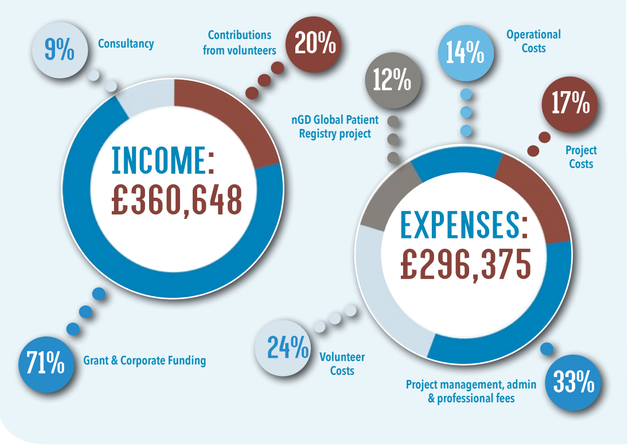
Dr Aimee Donald

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.



Our board of Directors, CEO, volunteers and doctors have donated over 2900 hours of their voluntary time to our activities.

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. No word can express your gift."

Youssef Mounib (Gaucher patient uncle from Morocco)

The work of the IGA Board of Directors is supported by very dedicated and passionate staff: Chief Executive Officer, Communications Manager, Finance Officer and Project Manager.

HUGE 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, Avrobio, Eurordis, ISU Abxis, Orphazyme; Pfizer, Sanofi Genzyme, Takeda.

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 Cheem 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.

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