

IMPACT REPORT 2020

Together we are stronger



INTRODUCTION:

The beginning of 2020 offered so much promised as we started the year with a visit to Pakistan to present and attend the 1st International Symposium on Gaucher disease, closely followed by the LDN WORLD meeting in Orlando, Florida where we presented two posters on our *Regional Manager Programme* and *A Global nGD Disease Patient Registry*.

However globally COVID-19 then dictated national lockdowns, shielding, anxiety and challenges to all of us never seen in our lifetimes. The IGA is extremely proud of its community that rallied around each other to offer guidance on clinical vulnerability, shielding, vaccinations, home therapy and sharing best practise of things that had worked in their country that may help others. Access to treatment and to clinical care/ advice was a challenge for our community and many patient had to miss their infusions. Clinical teams worked hard to resume treatment as soon as they could, to offer advice and provide reassurance using online technology, and support access to home therapy where possible.

We built our IGD 2020 campaign around *Homes Not Hospitals* to support access to home therapy for our community as this was a key challenge throughout the year for the community. We will continue to campaign for this in 2021 with the support of the EWGGD and other stakeholders.

Our plans for face to face meetings, projects and our biennial meeting were replaced by virtual meetings which although enabled us to continue to work on behalf of the Gaucher community and progress our programmes meant that some projects had to be re-shaped or postponed until a later date.

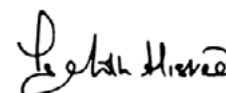
2020 saw our plans for GARDIAN, our patient led registry, progress as we finalised our nGD patient and observer reported outcomes and established a new company International GARDIAN Ltd (IGL), wholly owned by the IGA that will own and govern the registry.

Despite the challenges faced in 2020 patients still needed us to support them to access treatment, this area of the IGA's work is and will always remain a priority.

2020 was a huge challenge but as a community we were there for each other and I hope that our IGD slogan of 'Rare but not alone' has been evident to you all.



VESNA ALEKSOVSKA (CHAIR)



TANYA COLLIN-HISTED (CEO)

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 MISSION

to **empower** its members

to take **collective action** to address challenges Gaucher patients worldwide face in accessing **early diagnosis** and **optimal treatment and care**

To be the global voice for Gaucher patients and their families:

to **advocate** on behalf of Gaucher patients

to ensure that the Gaucher research agenda is **focused on patients' unmet needs**

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.



PATIENTS



DOCTORS &
HEALTHCARE
WORKERS



Our VALUES

Visionary

Patient

Passionate

We are:

Trustworthy

Collaborative



IN 2020 WE MADE THE DIFFERENCE BY:

- Helping to provide treatment to **31** patients in **11** countries including Afghanistan, Honduras, India, Jordan, Kenya, Morocco, Mozambique, Pakistan, Senegal, South Africa, Sudan.
- Hosting **7** educational webinars (town hall meetings) attended by **400** participants covering topics on: home therapy; comorbidities; gene therapy; digital health technology, clinical guidelines and newborn screening. These webinars were recorded and are available on our **Youtube channel**.
- Using International Gaucher Day to advocate for home therapy – on 1 October we were able to reach almost **4000** people on Twitter; almost **2500** people on Instagram and over **4000** people on Facebook.
- Becoming a registered charity. Since its inception, the IGA has been a not-for-profit organisation. This new charity status recognises the valuable work that the IGA does and demonstrates our commitment and values as we continue to serve the needs of our global community.
- Driving forward the development of GARDIAN – a global disease registry for neuronopathic Gaucher disease working in partnership with Kantar Health and Aparito, to be launched in 2021.
- Presenting at more than **6** virtual international symposiums and conferences on different subjects reaching an audience of doctors, researchers, nurses, patient advocates and patients on topics including the IGA and its global work; Gaucher disease and COVID-19; Quality of

Life measures; patient and specialist engagement: working together to improve healthcare for Gaucher; patient advocacy journey.

- Invited to attend and contribute in meetings, including: WORLD, ECRD, Round Table in Brussels, EWGGD, Council of European Federations; Rare Diseases International Meeting of Federations.
- Collaborating with pharmaceutical companies and sitting on working groups, advisory boards and project teams to benefit the Gaucher community: embedding decision making in healthcare white paper (Accea); patient representative on the Gaucher Outcome Survey (GOS) steering group (Takeda); member of the patient engagement panel advisory council (PEPAC) (Takeda); member of the Charitable access programme MEC (Takeda); Rare 2030 working group of experts; Drug Information Transparency & Access Task Force (DITA); EWGGD Board member.
- Our CEO and Chair visited Pakistan with Prof Timothy Cox to attend the 1st International Symposium on Gaucher disease hosted by Prof Huma Cheema. They also participated in faculty meetings at the hospital, a patient group



meeting, met the Minister for Health for Punjab and attended a press conference to raise awareness of genetic diseases in children in Lahore, its impact and the need for a rare disease policy and the development of a research unit.

- Providing evidence to the Ministry of Health in Ukraine on the procurement tenders for treatments for rare diseases, including Gaucher disease to express patient's great anxiety on switching of treatments annually. By working with the EWGGD we are seeking to develop a statement that would highlight the challenges, backed up with research and data on this approach to help treating physicians, patients and patient groups to have discussions with governments who are thinking about adopting this approach.
- The IGA co-authored/was involved in the publications of the following reports, papers and posters:
 - » ***The road to biosimilars in rare diseases - ongoing lessons from Gaucher disease***, published in March 2020 issue of *American Journal of Hematology* (Am J Hematol. 2020; 95:233-237): Guillermo Drelichman, Gilberto Castañeda-Hernández, Muhlis Cem Ar, Marta Dragosky, Ricardo Garcia, Howard Lee, Sergey Moiseev, Majid Naderi, Hanna Rosenbaum, Irena Žnidar, Andrés Felipe Zuluaga, Selena Freisens, Pramod K. Mistry.
 - » As biosimilars become a 'hot topic' in the rare disease world, the IGA started to educate its members about them at its biennial meeting in 2018 and continues to share more information in its newsletters. This

paper is of particular interest to doctors and patients who have encountered the following medicines: Abcertin, Asbroder and Glurazyme.

» ***The definition of neuronopathic Gaucher disease***. Journal of Inherited Metabolic Disease (Internet). 2020 Apr 3: Schiffmann R, Seigny J, Rolfs A, Davies EH, Goker-Alpan O, Abdelwahab M, et al. These are exciting times for our neuronopathic community with so much interest in research and medicines development. This new definition will help patients and clinicians with diagnosis and ultimately ensure that patients with nGD are able to access new medicines and the right clinical care.

» Two posters ***Regional Manager Programme*** and ***A Global nGD Disease Patient Registry*** were presented at WORLD symposium, EWGGD congress and ECRD summit.

- Presentations given at the EWGGD virtual congress: ***Our older generation, Development of the nGD PRO for GARDIAN*** co-presented with Elin Haf Davies; ***Regional Manager Programme*** co-presented with Dr Ashok Velodi and we also highlighted collaborative projects, including the development of the GD1 guidelines with Prof Chris Hendrickz.
- Working in collaboration with the European Haematology Association, we circulated a patient survey to help understand the impact of COVID-19 on Gaucher patients, their families and supporters and to provide invaluable research data with the intention of benefiting Gaucher patients. The results were presented as an e-poster at the LDN WORLD meeting 2021.

KEY ACTIVITIES:

CHARITABLE ACCESS

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 2020, we received requests for treatment from **11** countries such as Afghanistan, Honduras, India, Jordan, Kenya, Morocco, Mozambique, Pakistan, Senegal, South Africa, Sudan. By working closely with Sanofi Genzyme, Pfizer and Takeda we helped to give a future to **31** Gaucher patients as they received donated ERT. Sadly, **15** patients who reached out to us for support died without getting access to the treatment they needed. At the end of 2020 we have **13** cases still unresolved on our database.

One of the challenges we face in supporting the community is that there is very little access available to adult patients through charitable access programmes. This is an area we need to address in collaboration with our members and other stakeholders.

Sadly, there are many more other patients suffering without treatment across the world and therefore the IGA will continue to provide the much-needed support.



Jana (Jordan)



Nazlin (Kenya)



Shawn (Kenya)

COVID-19

We hosted a webinar with the EWGGD to learn more about the effects of coronavirus in Gaucher disease and to coordinate data collection, attended by over 100 participants including physicians, EWGGD colleagues, patients and patient advocates and industry representatives. Situational reports from countries across Europe were given and patient concerns were discussed. The IGA and pharmaceutical representatives were given the opportunity to speak, and questions were raised by patients and patient advocates. Throughout the pandemic we have continued to share best practices and updates, offering support to patient groups where needed.

One of the challenges that the COVID-19 situation has brought to Gaucher patients is receiving ERT infusions. The EWGGD published a statement recommending home treatment wherever possible and a webinar was hosted in May led by patient group representatives from Italy and Greece who have both been successful in lobbying their governments to authorise home therapy. The purpose of this meeting was to share best practice regarding home therapy and answer additional questions.



Home infusions for Aimee-Kate



Jade & her nurse Joey

INTERNATIONAL GAUCHER DAY

Our 2020 campaign focussed on the challenges Gaucher patients are facing during the COVID-19 pandemic and our aim was to improve patients' quality of life by advocating for home therapy.

On 1st October we were able to reach almost **4000** people on Twitter; almost **2500** people on Instagram and over **4000** people on Facebook.

We hosted a webinar which was attended by **60** participants from around the world and included an interview with a GD patient and her partner conducted by a clinical nurse from the UK. Our keynote speaker was Prof Derralynn Hughes who presented the EWGGD's statement on home therapy and we were once again joined by two patient group representatives from Italy and Greece who shared their experiences of advocating for home therapy in their countries. The video has also been viewed over **120** times from our YouTube channel.



Insta story infographics



INTRODUCING GARDIAN:

Gaucher Registry for Development, Innovation and Analysis of Neuronopathic disease (GARDIAN)

Neuronopathic Gaucher disease (nGD) has a high unmet need, but with an increasing number of pharmaceutical companies now seeking to develop potential investigational treatment options; it brings hope to many patients & families.

2020 saw a step forward in the development of GARDIAN:

- We developed a new nGD specific PRO (patient reported outcome) and ObsRO (observer reported outcome), that will be validated through the registry and owned by the IGA.
- We held an advisory board of key opinion leaders in the nGD world to advise on clinical data collection.
- We established a new company called International GARDIAN Ltd (IGL), owned by the IGA to govern GARDIAN.
- We appointed a Board of Directors to the IGL including representatives of; caregivers, clinicians, business (pharma and non-pharma), and patient advocacy.
- We secured funding to establish phase one of GARDIAN that will provide a non-site-based patients and carers reported outcomes and the specific nGD PRO and ObsRO that will be translated into 5 languages that will be accessible via an app or website.



EDUCATIONAL WEBINARS:

The IGA continues to support its members to boost their capacities through education and providing them with information about all aspects of Gaucher disease.

Following an assessment of training needs, we identified topics that were of importance for our community and during September and October we offered a series of online educational webinars.

These meetings were primarily for Gaucher patients and families but were open to the wider Gaucher community including clinicians and pharma representatives.

The webinars were conducted in a 'town hall' format which meant that there was lots of time for discussion and for participants to ask questions and share their experiences.

- **Gaucher & Parkinson's disease:** We were joined by three panellists: Prof Ari Zimran, Prof Per Svenningsson and Dr Marco Baptista who gave presentations and discussed this key topic. Margaret Giuliani from France moderated the session.
- **EWGGD GD1 guidelines:** The first joint EWGGD/IGA event to develop international type I Gaucher disease clinical management guidelines. Dr Derrallynn Hughes and Prof Magy Abdel Wahab hosted the meeting. Prof Chris Hendriksz presented challenges with diagnosis and clinical management in Africa.
- **IGD home therapy:** The focus of the IGD2020 campaign was home therapy, given many challenges that patients face when they had to go to a hospital, especially due to pandemic. We were joined by a clinical nurse, a patient and a carer who all shared their experiences of home therapy. There was also the opportunity to ask questions of Prof Derrallynn Hughes from the EWGGD and two patient group representatives who had advocated for home therapy in their countries.

CLINICAL GUIDELINES FOR GAUCHER DISEASE TYPE 1:

- **Ex vivo gene therapy for type I Gaucher disease:** Focused on the developments in AVROBIO's lentiviral gene therapy. AVROBIO's Chief Scientific Officer, Chris Mason, was our key speaker and Prof Timothy Cox from Addenbrooke's moderated this session.
- **AAV based gene therapy:** Focused on AAV-based gene therapy and Prevail's approach for neuronopathic Gaucher disease. Eriene Wasef, Medical Director at Prevail was our key speaker.
- All webinars were recorded and available on YouTube and on the IGA website.

This is a joint project with the EWGGD which started in September 2020. The European Working Group on Gaucher Disease (EWGGD) is a non-profit network established 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.

During EWGGD meeting (October 2020), four working groups were established:

1. Diagnosis,
2. Treatment and monitoring,
3. Comorbidities,
4. Outcomes that are important to patients and universal issues related to GD.

These working groups consist of physicians, researchers and patient representatives. There are several sub-groups within each working group, e.g. there are four sub-groups in WG4:

- 4.1 Supportive and symptomatic care
- 4.2 Care coordination and transition
- 4.3 Measuring outcomes
- 4.4 Self-management

The members of each sub-group/working group have regular meetings and they prepare the guidelines concerning the topic each group covers, and that will contribute to the international patient-centric guidelines for GD1 which will be published on the EWGGD's website.



Infographics from educational webinars

BIENNIAL ONLINE MEMBERS MEETING:

Due to the situation with COVID-19 pandemic, the IGA was not able to convene a face-to-face members meeting in 2020.

However, we recognize the importance of regular contact and meetings with our membership to enable us to take action on our work program, hear their needs and to share information and experiences and so we organised online members meetings.

The IGA online General Meeting was held in May and was a formal meeting with office and board reports being presented. The new IGA board which was elected came into office after this meeting. After the formal meeting, Prof Pramod Mistry and Tanya Colin-Histed gave an update on ongoing clinical trials and Dr Derrallynn Hughes updated on EWGGD, including COVID-19.

Following the online EWGGD Congress in October, we held a series of online members meeting. These took place on Mondays and Thursdays during the two last weeks of October. On Mondays, the meetings were open for our members only and the IGA board and staff team presented key projects and activities. Representatives from Canada, Japan, Romania, South Africa, Turkey, USA, Mongolia and Israel presented their work, shared experiences, successes but also challenges and concerns.

This session was very important and there was a lot of support, understanding, new ideas and good vibes. There was also a very interesting presentation from Orphazyme, giving an update on Arimoclomol.

On Thursdays we held educational sessions which our members had shown an interest in learning more about: Newborn screening (speakers Dr Juanita Navarette, Mexico and Dylan Simon, USA, moderated by Aviva Rosenberg) and Digital health technology (Daniel Lewi, Aparito, Michael Shapiro -Barr and Kathleen Coolidge, Backpack Health and Shelby Chamberlain, Patient Discovery). These meetings were open for all interested attendees and were recorded and available on YouTube.

All sessions were translated in real-time to Spanish to reach a wider audience.

**BIENNIAL
ONLINE
MEMBERS
MEETING**

**IGA INTERNATIONAL
GAUCHER ALLIANCE**
UNION OF GAUCHER PATIENT ASSOCIATIONS

Our members share best practice and highlight challenges: coming together as a community

A presentation from our member associations, including:

- funding for treatment
- communication strategies
- hosting events
- working with ministries of health

Monday 26th October

3 - 5.30pm (UK time)

gaucheralliance.org/events

Twitter, Instagram, Facebook, LinkedIn, YouTube icons

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.

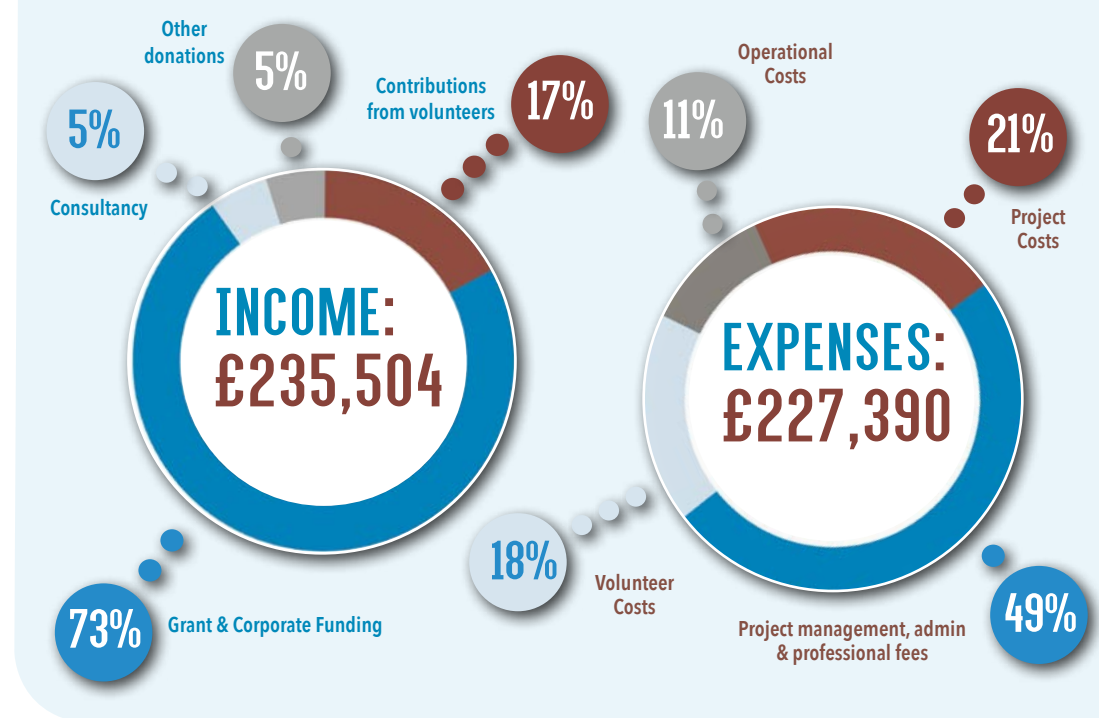
To ensure its independence, the IGA will only accept funds up to an amount not exceeding 35% of its annual budget from any one pharmaceutical company.

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 **1631** hours of their voluntary time to our activities.*

**IT IS EQUAL TO
£40,775**

We are extremely grateful for this dedicated support.



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!

ALEXION

AVROBIO

CENTOGENE
THE RARE DISEASE COMPANY

EURORDIS
RARE DISEASES EUROPE

ORPHAZYME

Pfizer

Prevail

SANOFI GENZYME

Takeda

Alexion, Avrobio, Centogene, Eurordis,
Orphazyme, Pfizer, Prevail, Sanofi Genzyme, Takeda.

IN 2021 WE INTEND TO:

- Collect data from the global Gaucher patient community on access to clinical care, diagnostics and disease management to 'map' local, regional and national challenges.
- Improve access to information through the translation of key educational documents and webinars.
- Use International Gaucher Day to focus on improving the diagnostic journey for Gaucher patients and by using patient stories to develop a series of key messages (red flags) on important symptoms that are potentially indicative of GD.
- Create three small focus groups representative of the Gaucher community in types, age and gender to develop a questionnaire that will seek to understand how the Gaucher community feel about gene therapy.
- Launch phase one of GARDIAN: We will undertake cognitive interviews to finalise the PRO and ObsRO, in six languages. The PRO and ObsRO will then be uploaded into GARDIAN and we will begin recruitment of patients and caregivers in mid 2021.
- Convene a face to face or series of virtual members meeting.
- Continue to build the capacities of the IGA membership by providing an educational webinar programme to include topics identified by our members as being of current interest to the wider Gaucher community, e.g. wellness for Gaucher disease; bone disease; biomarkers; fundraising; neuropathic GD and chaperone therapy.

- Actively recruit new volunteers through a structured programme to increase the resources available to the IGA to deliver more programmes and services to our growing community.
- Bring together a group of young adult Gaucher patients for a series of virtual meetings to identify current challenges and to review the Go With Gaucher (GWG) programme for 2022.
- Recruit a new Regional Manager for Africa as part of our ever-growing Regional Manager programme.
- In February 2021, to mark International Rare Disease Day, the IGA's Regional Managers will be conducting CMEs and other virtual events in different parts of the world to raise awareness about the clinical diagnosis, management and treatment of Gaucher disease. There are also plans to conduct physical awareness events in other regions later in the year if the situation with the pandemic permits.

International Gaucher Alliance

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