IGA INTERNATIONAL GAUCHER ALLIANCE

Driven by passion for patients

International advocacy for Gaucher patients since 1994

IMPACT REPORT 2023

Together we are stronger



INTRODUCTION:

Dear Friends

The International Gaucher Alliance (IGA) Impact Report 2023 reflects our dedication to advancing awareness, research, and support initiatives to enhance the lives of individuals and families affected by Gaucher disease.

Founded in 1994, the International Gaucher Alliance has been at the forefront of advocating for Gaucher community worldwide.

In the pages of this report, you will find a detailed insight into the International Gaucher Alliance's impact across various key areas, including advocacy, education, and patient support. By delving into the specifics of our activities and achievements, you will gain a comprehensive understanding of the extent to which the IGA has made a difference in the Gaucher disease landscape in 2023.

In 2023, the IGA held a "Go With Gaucher" meeting with many lectures, workshops and educational sessions for young people living with the disease. This meeting ended with the creation of a Youth Council initiative, that will be launched in 2024.

The IGA Strategic Plan 2024-2029 was developed with our community and stakeholders and outlines our priorities for the next five years.

Educational empowerment remains an important part of our advocacy, exemplified by our insightful and impactful webinars. This year we had a series of webinars on the topics of Gene therapy clinical trials and clinical trials for new treatment options for GD3 community. These webinars were organised in different time zones worldwide, so there were separate webinars for each part of the world with interpretation into four languages relevant to each region.

A significant highlight in our calendar is the annual celebration of International Gaucher Day (IGD), a momentous occasion where our global community unites to raise awareness, share stories of resilience, and advocate for improved access to diagnosis and care. This year, the IGA launched a movie ***The Power of Knowledge in a Timely Diagnosis – Building a Better Future***, in which families and physicians in different parts of the world speak about their diagnostic journeys to raise awareness of the importance of the power of knowledge in a timely diagnosis.

In 2024 the Regional Manager programme becomes the Global Gaucher Connect Programme (GGCP) and our Regional Managers are now Gaucher Leaders (GL). The GLs are the eyes and ears of the IGA and are responsible for working closely with key stakeholders in the Gaucher community such as patients, patient associations, doctors, industry, and policymakers; exploring and identifying new development opportunities; and identifying and addressing challenges faced by Gaucher patients in the region.

Integral to our success is the dedication of the participants in our volunteer program, passionate individuals who tirelessly contribute their time, skills and compassion to better the lives of those affected by Gaucher disease. As we reflect on the milestones of 2023, we express our deepest gratitude to our dedicated volunteers, whose passion and hard work have elevated the International Gaucher Alliance to new heights. Together, we continue to pave the way for a brighter future for Gaucher patients and families.

A final note is to reflect on the ongoing challenges faced globally by our community with patients and families displaced by war, or where there are a lack of access to treatment and care through political changes and challenges, as well as a lack of awareness of the needs of our community. The IGA has worked and will continue to work tirelessly for these patients and their families wherever they are in the world.

BILJANA JOVANOVIC (CHAIR) TANYA COLLIN-HISTED (CEO)

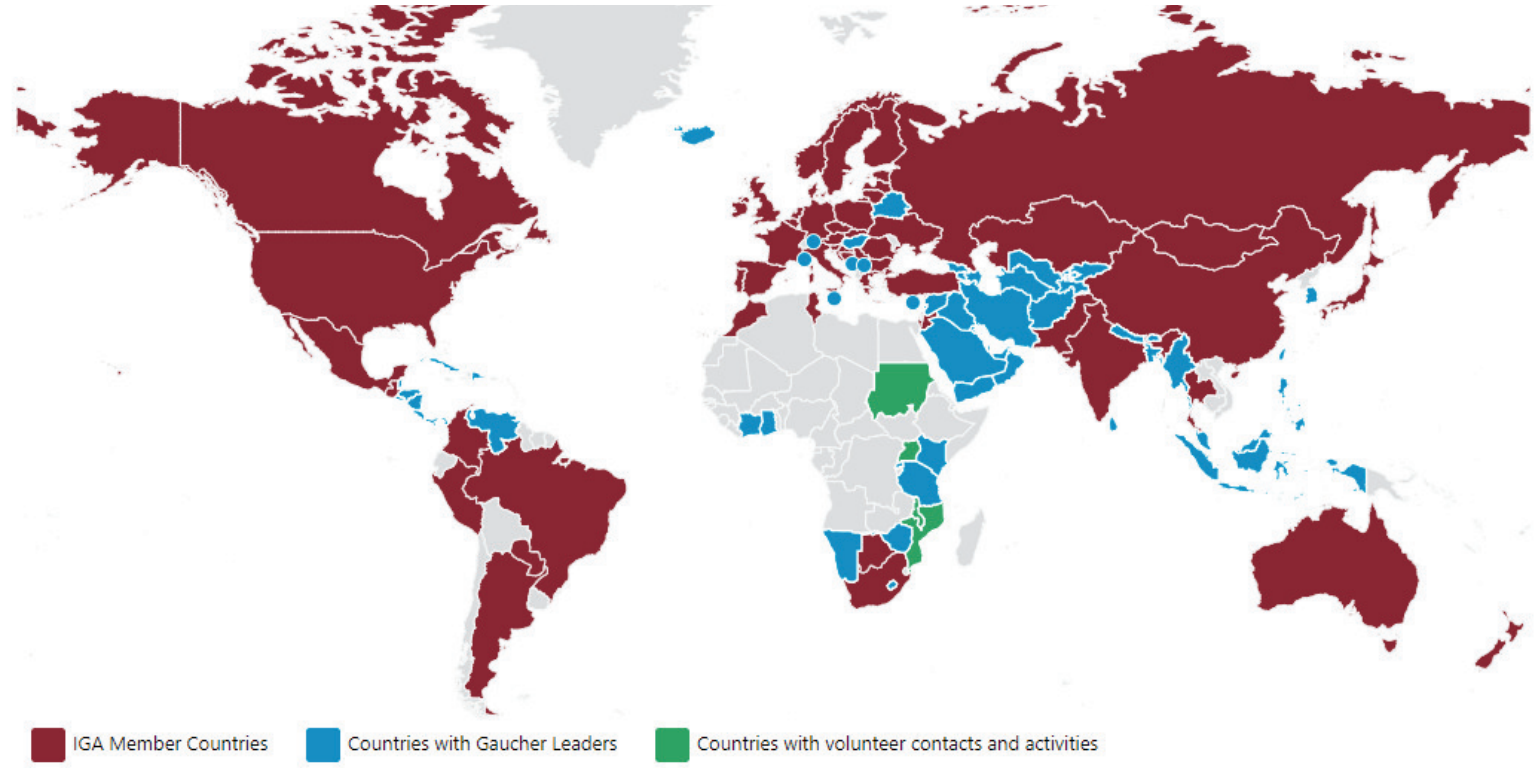
THE IGA:

The IGA is a patient-led international organisation that has become the 'go to' global voice for over **90%** of the Gaucher community (see map) 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 Global Reach







HOW WE MADE A DIFFERENCE IN 2023 :

- In 2023 the IGA was supported by 79 volunteers. We have been recruiting further volunteers to work on new projects. Volunteers came from 47 different countries (Argentina, Australia, Belgium, Botswana, Brazil, Bulgaria, Canada, China, Croatia, Czechia, Denmark, Egypt, France, Greece, Guatemala, India, Italy, Israel, Japan, Kenya, Lebanon, Lithuania, Malawi, Mexico, Mozambique, Nepal, Netherlands, Nicaragua, North Macedonia, Pakistan, Peru, Poland, Portugal, Qatar, Romania, Russia, Rwanda, Serbia, Slovenia, South Africa, Sudan, Sweden, Turkey, Uganda, United Kingdom, USA, Zimbabwe) and were involved in around 12 projects.
- We issued a non-comparable position statement in June (tiny.cc/psncm). The position statement outlines the IGA's stance in relation to the use of 'intended copies' of enzyme replacement therapy (ERT) medicines as a treatment for Gaucher disease, advocating for safe and effectiv. These 'copies' are also known as non-comparables (the term used in this statement), due to the fact that they have not been directly compared to the original medicine in a clinical trial setting.
- In October the International Working Group in Gaucher Disease (IWGGD) and IGA issued a consensus document on switching and monitoring best practice (tiny.cc/sambp). Born from concerns raised with the IGA by patients and patient organisations regarding being faced with switching enzyme replacement therapy due to payers' decisions around the procurement of treatment, the consensus document lists the points that decision-makers should consider prior to approving a new medicine purely on economic grounds
- We celebrated International Gaucher Day (IGD) with theme of **The Power of Knowledge in a Timely Diagnosis** producing a film in which patients and their families in different parts of the world speak about their challenges with diagnosis.
- We improved the visibility of the IGA website using search engine optimisation and other tools, ensuring that the IGA site ranks highly when people search for terms related to Gaucher disease. For example, we are the first result on Google when searching for charitable access to Gaucher medicines.
- The IGA operates a database where we record all requests we receive for charitable aid, and we advocate on behalf of the patient. The IGA has built up a strong relationship based on respect and trust will all of the companies and works very hard to support patients in a transparent manner. Visit tiny.cc/igacam for more details. The IGA received 17 formal requests for assistance to support access to charitable treatment through the IGA's online portal. These came from Cuba, Gaza, Botswana, Ethiopia, India, Kenya, Mongolia, Nepal, Philippines, Rwanda, Singapore, Uganda, Venezuela and Zambia.
- The IGA worked to re-organise and rebrand its Regional Manager (RM) programme ahead of a relaunch in 2024. The RMs improved awareness of Gaucher disease, including finding participants for the IGD video and co-ordinating local filming, as well as organising related events, including in Nepal and Botswana. They organised educational webinars for Africa, Asia, Europe and Central America, attended by over 2000 people (most of them medical professionals). RMs were in regular communication with medical professionals, researchers, volunteers and patient organizations to raise awareness of the work of the IGA. They also helped with the translation of materials which are shared in hospitals, doctors offices, conferences and other places where the Gaucher community gathers.
- The IGA continued to organise educational sessions in order to support Gaucher community to increase their capacities. These included:
    - *Substrate reduction therapy: What we need to know?* held for the Balkans region in Croatian, with interpretation to English.
    - Webinars on Gene therapy for Europe, Asia and the Americas were held in different time zones and interpreted into various languages.
    - Webinars on the GD3 Venglustat trial. Three webinars were held in different time zones for European, Asian and Americas region, also interpreted into various languages.

These webinars are available to view at tiny.cc/igavideos.

The IGA participated in the following working groups:

- Avrobio's GD3 Advisory Board to educate Avrobio on the GD3 global community to support the development of their GD3 GT clinical trial.
- Sanofi's GD3 Burden of Illness study – member of the Expert Review Group
- Member of the HTAi Rare Disease Interest Group and participating in:
  - WG 2 'Characterising Rare Diseases'
  - WG 3 'New paradigms for evidence generation and interpretation that takes into account the complexity of rare diseases including relevant value assessment frameworks'.
- Member of Eurordis Patient Advisory Board for the Screen4Care which offers an innovative research approach to accelerate rare disease diagnosis, based on two central pillars: genetic newborn screening and digital technologies. rebrand.ly/nbscreen)
- Member of GARDaccess (gardaccess.org)
- Member of RDI 'Identifying the socio-economic burden of rare disease in low middle income countries' project.
- On the advisory board of the European Haematology Association (EHA)
- Member of the RDI Global Access working group
- IWGD working group on Epilepsy in GD3
- IWGGD working Group on the use of Ambroxol for Gaucher patients, developing a best practice paper.
- Member of Eurordis Digital Advisory Group
- Member of the Gaucher Outcome Survey (GOS) steering committee for Takeda
- Attendance at the monthly C-Path for Lysosomal Disorders consortium meetings
- Member of the Medical Expert Committee (MEC) - Takeda's charitable Access programme.
- Member of the Mendelian Patient Involvement Initiative is a pilot project funded by an AI award by the National Institute of Health and Care Research (NIHR) that looks at how data centric tech could improver patients' lives by accelerating rare disease diagnosis and enabling faster, more targeted, access to treatments and management.

The IGA took part in the following meetings:

- WORLD Symposium, Jan 2023, Orlando Florida.
- The 3rd International Conference on Lysosomal Diseases (Med-Lysosomal2023), April, London – Presentation topic:
  - 'Recruiting patients for Gene Therapy clinical trials', is very concerning.
- Sanofi's Gaucher Leadership Forum, June Copenhagen – Presentation topic:
  - Gaucher Disease, an advanced but unfinished progress. Setting the path to accelerate Gaucher and ASMD patients' journey.
- IWGGD 10 May – Presentation topic:
  - IGA Position Statement on Non-Comparables
- HTAi Adelaide, Australia June – Presentation topics:
  - Rare Disorders. Is the Lack of Effect in a Patient-Reported Outcome, Reflecting no Benefit?
  - Developing a HTAi Rare Diseases Interest Group (RDIG) - Patients' Perspective
- Biosimilar medicines conference meeting, May Presentation topic:
  - 'Gaucher disease and remaining challenges.
- Sanofi's Rare Disease Registries Patient Council
  - Presented a poster: 'A rare partnership: community and industry collaboration to shape the impact of real-world evidence on the rare disease ecosystem.'
- The Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium, Jerusalem. Presented the above poster.
- The Regional Gaucher Meeting, Zagreb, organised by Croatian, Serbian, North Macedonian and Slovenian Gaucher Associations.
- Eurordis General Assembly – 17 May
- 2nd Symposium Recent Advances & Rare Overview, 26-27 February, Ponte Delgada, Portugal
- EURORDIS Council of European Federations Meeting – 12-13 December, Brussels Belgium.

KEY ACTIVITIES:

CHARITABLE ACCESS TO MEDICINES

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.

There were direct requests received for support to access treatment to IGA from 17 countries: Botswana, Cuba, Gaza, Ethiopia, India, Kenya, Mongolia, Nepal, Philippines, Rwanda, Singapore, Uganda, Venezuela and Zambia.

- 9 requests were approved
- 1 patient died before treatment could be found
- 9 cases remained unresolved: Jordan (4), Nepal (1), Afghanistan (1), Botswana (2), Singapore (1)

IMPACT OF WAR

In Sudan Gaucher patients and their families have been displaced, treatment in the country destroyed. Avenues to get treatment into Sudan remain very challenging and impossible for periods of time. Sadly many Gaucher patients have died and others have been without treatment for months. Some patients have managed to travel to other countries to access treatment and clinical care. The IGA in partnership with Direct Relief (the distributor for the Takeda charitable access programme) and Sanofi have worked closely together on this with clinicians and patients/families.

In Gaza communication with patients has broken down and a new patient planning to be seen at the Shaare Zedek hospital in Israel for support has not been able to.

Some patients remain in Ukraine and are receiving treatment and care, others have gone to other countries. In late 2023 a new patient with suspected Gaucher disease approached the IGA for help and together with Dr Andrea Dardis, Italy, this patient is being supported.

OTHER SUPPORT TO PATIENTS AND FAMILIES

Requests for support were received from Sweden, Peru, Iran, Egypt, Pakistan, Sudan, Ukraine, South Africa, Morocco, Turkey, India and France and included:

- Relocation and link to clincial expert
- Relocation and access to medicines
- Access to diagnostic services
- Prescribed medicines not arriving in country.
- Access to care and medicines through war displacement
- Support for further information into different disease issues and links to experts
- Challenges with access to medicines through Government budget reductions
- Issues with Insurance coverage for medicines

GENE THERAPY

In 2022 the IGA made a commitment to educate the international Gaucher community on Gene Therapy and embarked on a series of activities, in 2023 we achieved the following.

- Manuscript: January 2023 saw the publication in Orphanet of the IGA's international survey that sought to understand patient and parent and caregiver perceptions on gene therapy in Gaucher disease (insert link to paper).
- Website: Launched a gene therapy web page to enable the community to be able to easily understand the similarities and differences in the current clinical trails for Type 1 Gaucher disease (tiny.cc/gtct).
- Educational webinars: hosted a series of regional website on the current clinical trials for Type 1 Gaucher disease to ensure equity of access in different time zones and languages. These webinars are available on the IGA's YouTube channel (tiny.cc/igavideos).
- Educational Awareness of Clinical trials: hosted a series of regional website on the current clinical trials for Type 3 Gaucher disease to ensure equity of access in different time zones and languages. These webinars are available on the IGA's YouTube channel (tiny.cc/igavideos).

STRATEGIC PLAN 2024-2029

In 2023, the IGA worked collaboratively with many stakeholders in the Gaucher community to develop its new Strategic Plan for the five years from 2024.

This plan sets out a bold new vision for the IGA's work, aiming to cement its reputation as the go-to international organisation representing and advocating for Gaucher patients and their carers.

Read the Plan at gaucheralliance.org/sp2429

INTERNATIONAL GAUCHER DAY



The Power of Knowledge in a Timely Diagnosis

The main event for International Gaucher Day (IGD) in 2023 was the video, ***The Power of Knowledge in a Timely Diagnosis – Building a Better Future.***

We shared the video on Facebook and Instagram. We also uploaded it to YouTube and shared it on LinkedIn. As of the end of 2023, the English version of the video had reached 1,600 people on Instagram, 2,000 on Facebook and had been viewed 264 times on YouTube.

The video subtitles were also translated into Spanish (released on 1 October), Serbian and Portuguese, the other two languages used in the video, followed later in the month, with Macedonian, French, Hindi, Chinese, Russian and Arabic added a few days later.

An accompanying leaflet for the video in Afrikaans, Arabic, English, Spanish, French, Croatian. Latvian, Macedonian, Ndebele, Portuguese, Russian, Shona, Tsonga, Xhosa and Zulu versions is available on the website.

We also made all of the social media posts available for download, including Word templates that people could download and use with translated text.

We made fewer social media posts this year as we wanted the video to be the focus. We took quotes from each of the video participants to make the posts. We made a total of 14 Facebook posts. Total post reach was 3,780. Total engagement was 571. We also made 14 Instagram posts. Total post reach was 3,866. There were 821 likes. We made 13 LinkedIn posts. There were 3,180 impressions and 99 reactions.

Watch the video on our YouTube Channel: tiny.cc/igavideos





GARDIAN:

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

About GARDIAN

In 2020 the IGA set up a company called International GARDIAN Limited (IGL). IGL is wholly owned by the IGA, therefore the global Gaucher community will own and govern a global patient registry for Types 2 and 3 Gaucher disease.

GARDIAN will benefit patients and caregivers by increasing the understanding of the impact of the neurological manifestations of the disease, improve access to and achieve a timely diagnosis, inform better care and support, and enable better targeted research and the development of better safer treatments.

GARDIAN was launched on April 25th, 2022, and is available in 8 languages UK English, US English, Arabic, Japanese, Chinese, German, Spanish, and French.

- February saw the first GARDIAN multistakeholder meeting, bringing together 30 attendees from the registries scientific advisory board, pharma Industry working group and members of the IGL board of Trustees.

- July saw the publication in Orphanet of 'A global neuronopathic Gaucher disease registry (GARDIAN): a patient-led initiative' (tiny.cc/gardianorphanet).

- December 2023, the Gaucher Community Alliance, and IGA teamed up to host a virtual focus group, featuring 12 participants, including parents, caregivers, and individuals living with neuronopathic Gaucher disease (nGD. The session aimed to explore the awareness and utilization of the GARDIAN registry platform, and to develop strategies to enhance participation among the target demographic.

- Participated in a two-day workshop on establishing a community governed patient registry and offered learnings from GARDIAN. The workshop was hosted by European Health Data & Evidence Network (EHDEN), a member of the GARDIAN team.







GARDIAN was presented at various meetings and conferences during 2023, including:

Poster

- WORLD 2023 poster: GARDIAN Patient Involvement in the Development of Outcomes Measures Specific to Neuronopathic Gaucher Disease Type 2 and Type 3 to Assess HRQoL Over the Disease Journey.

Meetings

- IWGGD, May 2023, GARDIAN registry for GD2 and GD3
- European Human Genetic Society (EHSG), Glasgow, June 11-13, Presentation topic: Engaging patients in clinical trials endpoints: A Gaucher Disease Registry perspective

Go with Gaucher Istanbul

Taking forward the next generation

The IGA successfully hosted its fourth Go with Gaucher (GwG) conference in Istanbul, Türkiye, from November 10th to 12th.

The conference was specifically designed for young adult Gaucher patients aged 18-30, offering them a unique opportunity to connect with fellow patients worldwide and gain deeper insights into Gaucher through expert-led lectures on crucial topics.

Attended by over 15 patients representing Europe, the Americas, and Asia, the conference featured expert talks covering various pertinent subjects. These included recent advancements in Gaucher disease research and treatment, managing mental health during significant life transitions while coping with Gaucher disease, transitioning between different therapies, the significance of diet and nutrition for Gaucher patients, engaging in the Gaucher community through advocacy, and the impact of Gaucher disease on pregnancy.

Esteemed speakers such as Prof. Derralynn Hughes (Royal Free London NHS; Chair, IWGGD), Zubyda Azzam (Rareminds), Dr. Mehmet Balcı (Istanbul University), Gidon Goodman (Gaucher patient and President of the Gaucher Association of Australia and New Zealand), and Elizabeth Morris (Clinical Nurse Specialist, Cambridge University Hospital) shared their expertise. Moreover, participants courageously shared their poignant and inspiring personal narratives of living with Gaucher during the patient monologues session.

The event left a profound impact on the attendees, many of whom expressed how it was a life-changing experience for them.



"It was a wonderful experience to meet other Gaucher patients from literally all over the world. It is an incredible experience to share the feelings of the disease you have had for years with people from other geographies who experience the same feelings."

– Deniz Erdem, Türkiye

"Attending the Go with Gaucher conference in Istanbul was an enlightening experience that transcended borders. Engaging with experts from around the globe not only deepened my understanding of Gaucher disease but also fostered a global community committed to advancing research and support for those affected."

– Saurabh Kapse, India

"I had the great opportunity to travel to Istanbul, spend such wonderful, enriching days full of important information. So thank God and you, IGA, for the great help and opportunity provided."

– Emilse Peña, Guatemala.

Two Regional Managers report:

Patricia Lucki



2023 has been a busy year for IGA in Central America. We started projects of e-health and registries in Guatemala, while advertising GARDIAN. We worked with other associations in other countries, in rising awareness of rare diseases and the need of neonatal screening, including lysosomal diseases.

As we partner with CEPCAL, “Colaborativa para las Enfermedades Poco Frecuentes en el Caribe y América Latina”, we have been involved in the organisation of the 2024 Regional Congress, with a working group that will focus on Gaucher Disease in the region.

We also started the process of distributing leaflets about GD in paediatric congresses and emergencies of hospitals, while we participate as speaker in some of them.

Finally, we are doing our best effort to support patients in countries where there are no associations or where their migration or political conditions do not allow them to receive treatment. We look forward to seeing more progress in such a complex situation.

Andre Balzekiene

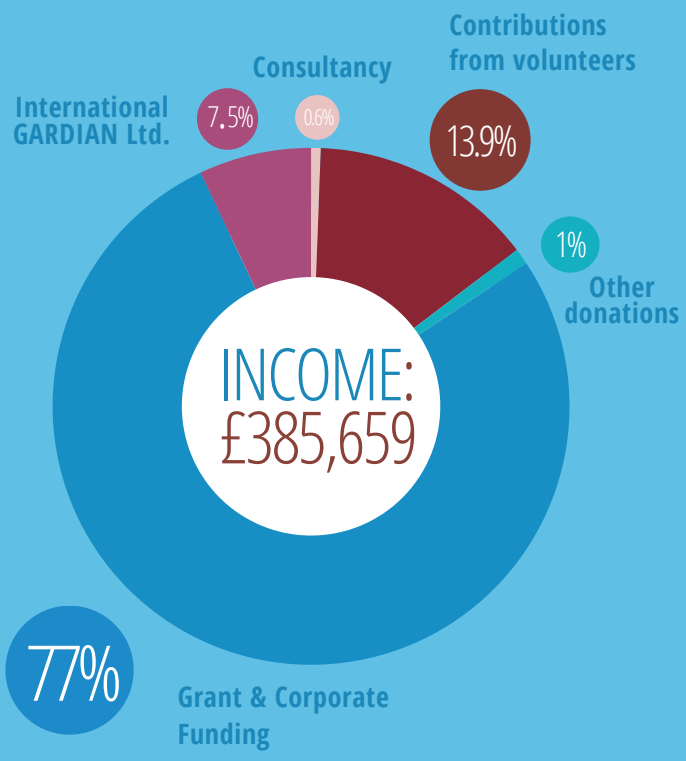


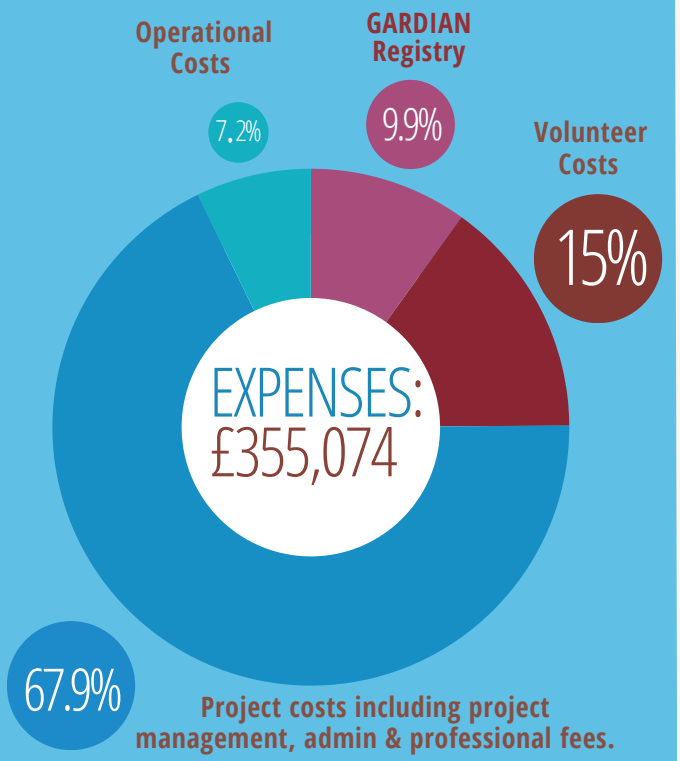
In 2023 I was the Regional Manager for Europe where my countries of interest vary from Iceland or Cyprus to Belarus or Uzbekistan. Since these countries do not have local Gaucher associations, my main goal is to find out if there are any Gaucher patients living there, and what are the laws and treatments for this or other rare diseases. Trying to reach out and find medical professionals or government officials to reply to your email often seems like a mission to the unknown land and I am very happy every time I receive a reply. I’ve made contact!

I also helped to organize a webinar for raising awareness on Gaucher disease among health professionals. It was my first job at the IGA and I was happy to be part of it.

The biggest highlight of 2023 was the first time opportunity to participate in a regional meeting of patients and medical professionals in Croatia. It was an amazing possibility to get to know other patients, meet the doctors, and discover the latest clinical data. The greatest thing on this volunteer journey is the family-like feeling you get along the way.

FINANCE:





Our board of Directors, CEO, volunteers and doctors have donated over 2156 hours of their voluntary time to our activities.

THIS IS EQUAL TO £53,431.

We are extremely grateful for this dedicated support.

The work of the IGA Board of Directors was supported by very dedicated and passionate staff: Chief Executive Officer, Operations Officer, Communications and Campaigns Officer and Projects Officer.

The IGA is funded through grants and sponsorship from a number of companies and organisations, and through our CEO and Board members undertaking consultancy work.

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.

A HUGE THANK YOU TO OUR SUPPORTERS...

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

Avrobio, EURORDIS, Freeline, Gain, Horizon, Lilly, Pfizer, Sanofi, Takeda.

The IGA Impact Report gives an overview of the IGA's finances for 2023 on a cash basis. This differs from our statutory accounts which are done on an accrual basis. Please keep this in mind if comparing the documents. Many of our projects are long term and therefore any unspent funding will be rolled into the following year.

IN 2024 WE INTEND TO:

- Celebrate 30 years of advocating on behalf of the Global Gaucher community with various events including a 'fire side chat' looking back over the past 30 years.
- Hold our biennial members meeting in November in Lisbon, Portugal and invite our Gaucher leaders and volunteers to join us.
- Launch the GARDIAN dashboard to share some of the data captured in the nGD patient reported registry.
- Continue our work with the older generation of Gaucher patients to better understand their clinical and non-medical needs and seek ways in which to address unmet needs where they exist.
- Understanding the community's position on Newborn Screening, providing a position paper that will be a tool for patients and advocates to address the issue in their own country.
- Provide a series of podcasts on different aspects of Gaucher disease e.g. gene therapy, pregnancy, switching to an oral therapy, transition, mental health.
- Continue to grow our Global Gaucher Connect (formerly Regional Manager programme) in Africa, Central America, Southeast Asia, Eastern Mediterranean and European regions.
- Seek to work in collaboration with other stakeholders to develop a patient registry in Africa.
- Continue to ensure access to information and our services by providing translation and interpretation in as many languages as possible.
- Through the 2023 Go with Gaucher symposium, to work with the young adults to develop a Youth Council for the IGA. This will create the opportunity to have young people involved in the IGA structure, and their perspective in the work of IGA.



The IGA Board and Staff