IGA INTERNATIONAL GAUCHER ALLIANCE

Driven by passion for patients

IMPACT REPORT 2022

Together we are stronger

GARDIAN

THE NEURONOPATHIC GAUCHER REGISTRY

OWNED AND GOVERNED BY THE GLOBAL PATIENT COMMUNITY

First global, patient-led registry for **Type 2** & **3 Gaucher Disease**

INTRODUCTION:

Welcome to our 2022 IGA Impact Report. First of all, we would like to congratulate and celebrate everyone for what has been achieved over the past 12 months for the Global Gaucher community. It is only as a community that we will be able to continue to improve patients' lives and take the steps forward needed to bring new knowledge, and new treatments, to patients and ensure equity for our community.

In 2022 we welcomed two new Members to the IGA with Peru and Argentina joining, making our membership 58 countries. However as you can see on the map on page 3, we have a good coverage of the globe through our programmes.

We managed to hold a Members' meeting and to meet IGA Members' representatives and volunteers face to face and virtually in Rotterdam, the Netherlands. It was wonderful experience to finally, after all challenges and difficulties, meet our dear friends who work along with us to achieve our goals.

We are constantly inspired by people's resilience and passion to fight for their child or give their time to help others; the Gaucher community is very good at this and our 'what we have achieved' section demonstrates this in statistics about our volunteers and new patients receiving ERT through charitable programmes. A lot of things in life are hard but as we have demonstrated not impossible.

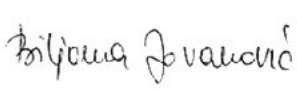
Special thanks must go to the IGA Board of Directors who are all volunteers and give their time tirelessly to improve patient outcomes, and break down barriers across the world, and to the International Working Group on Gaucher Disease (IWGGD) Board that also give their time to increase standards of care and gain a better understanding of unmet needs through working groups, webinars, and scientific meetings. The IGA office's support made all of these improvements possible through their hard work, enthusiasm and knowledge, and we thank them very much.

2022 saw new clinical trials for Gene Therapy, and with a number of different mechanisms it is important to ensure that the patient community truly understands Gene Therapy, its possible opportunities and challenges. The IGA is working collaboratively to develop an educational programme for our community.

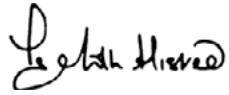
The focus of International Gaucher Day 2022 was the management of Gaucher Disease, focusing on the whole person beyond treatment for the physical symptoms.

Recognising our global role and responsibility, in 2022 we strived to provide translations of our work in many areas including webinars and during our biennial meetings and surveys; we commit to continue this and, going forward, will be providing more languages in partnership with our volunteer translators and Member organisations.

Enjoy our Impact Report. If you are not currently involved in our work, please do contact us – it is only together that we are stronger.



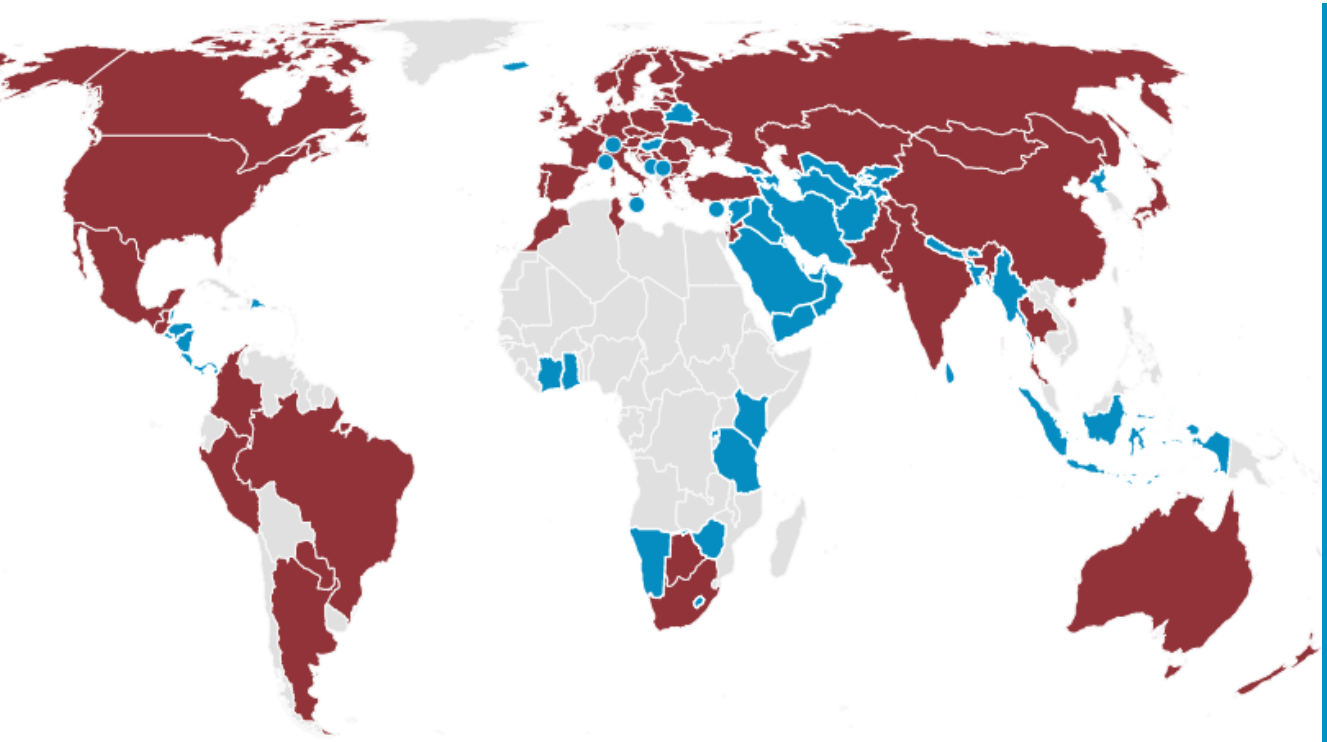
BILJANA JOVANOVIC (CHAIR)



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.



IGA Member countries

Countries with Regional Managers

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 VALUES

We are:

- Visionary
- Patient focused
- Passionate
- Trustworthy
- Collaborative

Our MISSION

PATIENTS

DOCTORS & HEALTHCARE WORKERS

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

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

IN 2022 WE MADE A DIFFERENCE BY:

- Launching the GARDIAN Registry, which is the first global, patient-led registry for Type 2 and 3 Gaucher disease. It will capture the burden of the disease on patients' everyday lives to build a picture of its natural history without a targeted medicine that addresses the neurological aspects of the disease.
- Holding our first in-person Biennial Members Meeting since the pandemic in Rotterdam, the Netherlands. This was an opportunity for members of the IGA to connect, give and receive updates on their work and the work of the IGA, take part in workshops and hear presentations from pharma companies and others working in the rare disease field.
- Expanding our volunteer programme to help us reach more people in different parts of the world, including by translating documents and supporting our regional managers.
- Helping 18 patients in India, Africa and the Middle East gain charitable access to life-saving medicines.
- Holding International Gaucher Day with theme of **Body Mind Soul** promoting a holistic approach to diagnosis and treatment.
- Welcoming two new members, in Peru and Argentina, bringing our membership total to 58.
- Launching a new website to help people find information about Gaucher disease and what the IGA offers more easily.

IGA volunteers made a difference by:

- At the end of 2022, 45 volunteers from 40 different countries were involved in over 11 projects (almost all IGA projects in 2022 had volunteers working on them). They collectively volunteered for 2083 hours in total. Our volunteers were recruited through advertisements on social media, conferences and events, and some of them were invited by other volunteers, staff or board members.
- Thanks to volunteers we were able to translate many materials into Arabic, Spanish, French, German, Japanese, Chinese – and with that to reach out to families and patients around the world.
- Volunteer activity also included the home therapy project, the older generation project, Regional Manager Programme, GD1 guidelines, the self-management working group, switching and monitoring of patients, non-comparable and biosimilar medicines, GARDIAN recruitment (through webinars, translation and sharing of materials), development of the new 2024-2029 IGA strategy, Scientific Advisory Committee and the development of neuronopathic guidelines for care and support.
- Reviewing and reforming the IGA Regional Manager (RM) Programme continued with the process of recruiting volunteers to support the RMs. The RMs were able to gather a lot of information from many different countries that will help us in the realisation of activities in 2023. Now we are equipped with the necessary to knowledge to move forward and do more in the countries where there are no patient groups or organisations.

The Regional Managers are the eyes and ears of the IGA and are responsible for working closely with key stakeholders 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. We ended the year with RMs in Southeast Asia, the European regions 1 and 2, the Central America 1 region, the Eastern Mediterranean region and the African region.

Participating in the following working groups:

- International Working Group on Gaucher Disease (IWGGD) – Supportive care
- Global Alliance for Rare Diseases (GARD)
- Eurordis Digital Advisory Group (DAG)
- Rare Disease International (RDI) Global Access Working Group
- Eurordis Screen4Care patient advisory Board
- Member of the Medical Expert Committee (MEC) – Takeda's Charitable Access Programme
- Gaucher Outcome Survey steering committee – Takeda

The IGA co-authored/was involved in the publications of the following reports, presentations, papers and posters:

- Transformative effect of a Humanitarian Program for individuals affected by rare diseases: building support systems and creating local expertise: Orphanet – IGA CEO co-author, publication
- Patient centered guidelines for the laboratory diagnosis of Gaucher disease type 1: Orphanet – Jasenka Wagner co-author, publication
- Findacure, now Beacon, February 2022: Building an international patient group: achieving a global reach with limited resources – poster
- Lysosomal Storage Disorders, A practical Guide, Atul B. Mehta and Bryan Winchester, 2nd Edition, 2022: Patients' Perspective; Chapter, pp.299-307 – IGA CEO co-author, publication
- European Conference on Rare Disease (ECRD): Harnessing the power of patients: Developing and validating the Neuronopathic Gaucher Disease Patient Reported Outcomes (nGD-PRO) and Observer Reported Outcomes (nGD-ObsRO) to measure HRQoL in patients with Gaucher Disease Type 2 and Type 3 – poster
- Creation of the GARDIAN patient registry for neuronopathic Gaucher Disease Type 2 and Type 3: A collaborative approach – poster
- Global Genes: Creation of the GARDIAN patient registry: A collaborative approach to address the unmet needs of patients with neuronopathic Gaucher disease type 2 and type 3 – poster
- Understanding patient and parent/caregiver perceptions on gene therapy in Gaucher disease: an international survey: Freeline, Pfizer, Avrobio.

ISPOR - The Professional Society for Health Economics and Outcomes Research:

- Addressing unmet needs of patients with neuronopathic Gaucher Disease Type 2 and Type 3: Creation of the GARDIAN patient registry – poster
- 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 – poster

International Working Group on Gaucher Disease biennial meeting:

- GARDIAN – presentation
- Procurement of treatment for Gaucher disease – presentation

KEY ACTIVITIES:

HUMANITARIAN AID

In 2022, the IGA received requests for assistance to access treatment from patients, family members and doctors in the following countries: Afghanistan, Cuba, Ethiopia, Ghana, India, Jordan, Kenya, Morocco, Mozambique, Peru, Qatar, Mongolia, Sudan, Venezuela, Palestine, Rwanda, and Zimbabwe.

Sadly, there are a number of cases that have still not been resolved; this is due to several different challenges, including:

- The country of residence is not covered by certain company programmes
- There is a maximum quota per country, and this has been reached
- There is a challenge with treating adults due to eligibility
- Access into the country is restricted due to war and/or political challenges
- The process of getting free treatment into a country is lengthy and challenging

This includes patients in Afghanistan, Ghana, India (12), Jordan (4), Peru, Venezuela, Zimbabwe (2), Mongolia

Approved – 18 patients: India (7), Kenya (2), Morocco (2), Mozambique , Qatar (2), Sudan (2), Rwanda, Palestine

Deaths – sadly two patients lost their lives. One was very ill, and treatment did not reach the patient in time. In this case, access to diagnosis and awareness was essential; in the other case the patient died of another cause unrelated to Gaucher disease and had been doing very well on treatment. These patients were from India and Ethiopia.

In process – These cases are being processed and we anticipate that these patients will have access to treatment in 2023. Cuba (1), India (2)

Ukraine – The IGA were in touch with the Gaucher patient group in Ukraine after the war started and supported the companies that were treating patients to ensure access was still available to patients, either in Ukraine or to where they were now living.

The IGA works collaboratively with the three pharmaceutical companies that provide charitable treatment to Gaucher patients.

These are:

- Takeda Charitable programme: through the Medical Expert Committee (MEC) programme (the IGA is a member of the MEC) in 2022 a total of 35 patients was approved from Ethiopia, Kenya, Pakistan, Tanzania, Tunisia, and Sudan.

- PfizerCares: seven patients approved with a further 3 in the pipeline (included in the figures above).

- Sanofi ICAP: 11 patients approved for treatment (included in the figures above). Please note that this figure only includes those patients referred to Sanofi by the IGA to their programme and therefore does not represent the total number of other Gaucher patients that were approved by ICAP through other channels. These patients were from India and Ethiopia.

NON-COMPARABLES

The IGA began work to agree a position on non-comparable medicines for Gaucher disease on an international scale, seeking to understand the opinions, experiences and thoughts of the worldwide Gaucher community to develop a statement that outlines our position clearly, factually and with confidence.

STRATEGIC PLAN

A working group was set up to review the IGA's Strategic Plan with a view to updating it for the next five years (2024-2029). This includes looking at whether we need to update our mission and objectives. We are consulting widely with various stakeholder groups.

NEW WEBSITE AND SYSTEMS

A new website was brought online, incorporating new systems to make communications easier, including new ways of collating Country Reports, to collect data from our Members and Regional Managers to give us a much better understanding of challenges, unmet need and access to care and treatment. We also improved the system for Humanitarian Aid requests.

VOLUNTEER PROGRAMME

Our new Projects Officer reorganised and streamlined the Volunteer Programme, bringing new people on board and ensuring that their time was used effectively, leading to a year-on-year increase in time volunteered, and increased output in areas such as translations.

SPOTLIGHT ON AN IGA VOLUNTEER

I decided to volunteer as Regional Manager for the IGA because I believe in the need for a more sustainable world and I cannot understand sustainability without solidarity. The Regional Manager programme allows organized Gaucher patients to reach those who have no local support or framework and this adds meaning to our own local work. I expect to reach, hand in hand with the IGA, underserved patients and create a network in the Central American Integration System (SICA) region.

In practical terms, this means to include in the regional health agenda rare diseases research, treatment and care and to have a comprehensive catalogue that includes Gaucher disease in the regional diseases classification; to improve regional registries; and to have clear policies regarding treatment acquisition and registration. This year we expect to start a virtual reference center and this will be the first step towards our final objectives.

Patricia Lucki

INTERNATIONAL GAUCHER DAY

Our 2022 campaign focused on a holistic approach to diagnosis and treatment with the theme Body Mind Soul Managing Your Gaucher Disease.

We shared the stories of patients from around the world, both in social media posts and in videos.

We also shared the views of clinicians and others working in the field.

We made a total of 37 Facebook posts and another on 27 Instagram. We sent 26 Tweets and made 16 LinkedIn posts. We also shared videos in different languages. Members and partners also posted, both original posts on the theme and using our templates, with some translating the messages into their languages.

IGA CEO CELEBRATES 25 YEARS

On Friday 18 November, the UK Gauchers Association and the IGA held receptions for IGA CEO Tanya Collin-Histed to celebrate her 25th anniversary of being a patient advocate for Gaucher patients around the world.

The UK GA event was a lunch attended by around 20 people, where Tanya was presented with an engraved fountain pen.

The IGA held a reception in the afternoon with people attending virtually from around the world as well as in person. Tanya received an engraved vase and a book of photos and reminiscences contributed by around 100 people from all over the world. Attendees including patients, carers, health care providers, researchers and representatives of pharmaceutical companies spoke, paying tribute to Tanya and her accomplishments over the years.

BIENNIAL MEETING:

On the weekend of 6-8 May, the IGA held its first Biennial Members meeting since the pandemic in Rotterdam in the Netherlands.

Over 40 people from around 30 countries, most in-person with a few online, attended. It was clear how happy people were to once again be able to meet face-to-face, something that hadn't happened since 2018.

This was an opportunity for Members of the IGA to connect, give and receive updates on their work and the work of the IGA, take part in workshops and hear presentations from pharma companies and others working in the rare disease field. Dr Lucy McKay from Medics for Rare Diseases gave a presentation on her organisation's work, and Barbara Sjouke MD PhD from the Amsterdam University Medical Centre spoke about gene therapy. Takeda, Sanofi, Freeline and Prevail shared the industry perspectives.

The meeting also gave recognition to the volunteers who serve the IGA in many capacities, including the Regional Managers and others who work so hard on our behalf.

Biennial meeting participants

SPOTLIGHT ON AN IGA VOLUNTEER

I'm a mother of two fortunate Gaucher patients. I volunteered at the IGA, determined to make a difference, and help those in need. One issue that can be resolved is the language barrier; spreading enlightenment and making everyone aware of available options conveyed in their language can make a huge difference and ensure that no one faces the consequences of ignorance. Thank you, IGA, for giving me this valuable chance to deploy my humanity.

Nuha Ahmed

GARDIAN:

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

GARDIAN was presented at various meetings and conferences during 2022:

- 3 webinars in Spanish (Peru, Argentina, Mexico), September – October 2022
- Gaucher family meeting, Italy, October 2022
- Gaucher family meeting, Canada, November 2022
- Central America forum on reference centers for rare diseases, November 2022
- LSD conference, Panama, October 2022
- Gaucher family meeting, Denmark, November 2022
- Gaucher Community Alliance family meeting, USA, October 2022
- Genetic Rare Immune Disorders Symposium, USA, October 2022
- Global Genes conference, September 2022
- ISPOR, November 2022
- Rare disease conference, Austria, December 2022
- Webinars for UK, Egypt, Argentina, Spain, Mexico, Peru, Japan

GARDIAN
THE GAUCHER NEURONOPATHIC REGISTRY
OWNED AND GOVERNED BY THE GLOBAL PATIENT COMMUNITY

To support the launch and take GARDIAN forward we have:

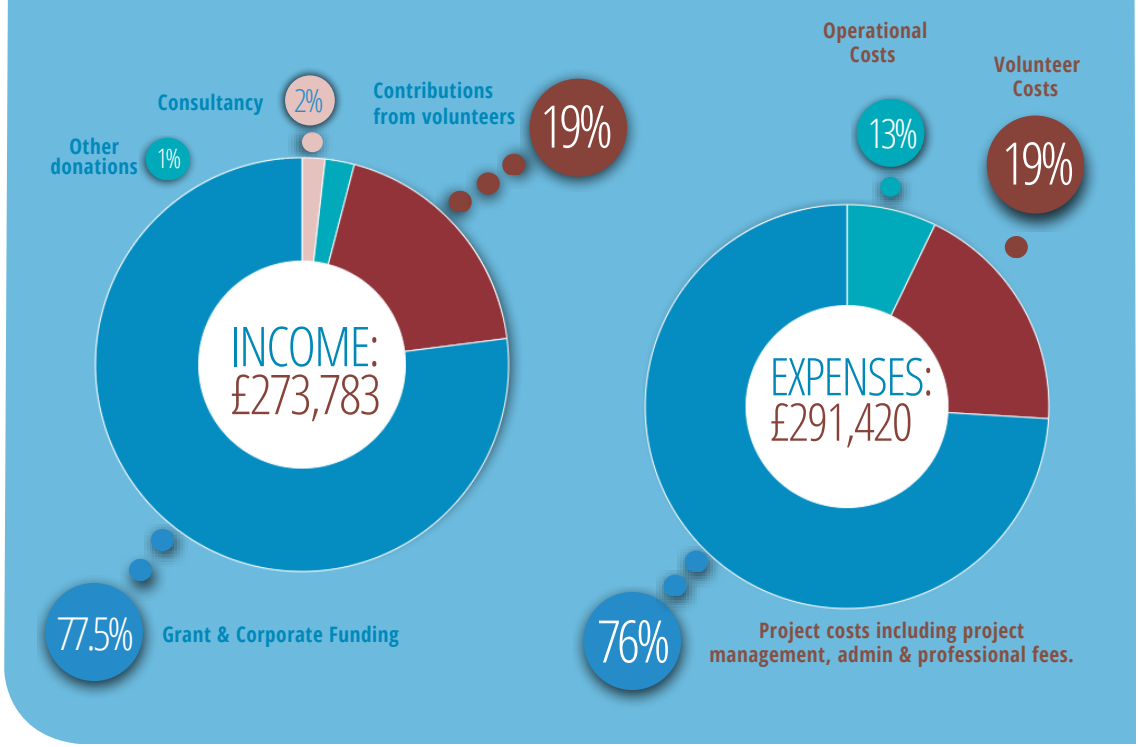
- Appointed Tanya Collin-Histed as CEO to support the work of the IGL Board.
- Formed a **Scientific Advisory Board (SAB)** to advise the Board of Directors of IGL on scientific and clinical matters, consider and approve research and data requests and contribute to the on-going development of GARDIAN for the benefit of the global patient community. The SAB met for the first time on the 29th September.
- Formed a **Pharmaceutical Industry Working Group** to regulate the relationship with pharmaceutical and biotech industries, define rules for sponsorship opportunities, define access and scope of access to non-identifiable, aggregated data stored and managed by GARDIAN and approve and/or partner with these organizations for scientific publication aligned with the main objectives of GARDIAN. The PIWG met for the first time on the 30th November and has a membership from 7 companies: Avrobio, Freeline, Gain, Pfizer, Prevail, Sanofi and Takeda.
- Formed the **Community Working Group** to:
    - Identify areas of research that GARDIAN could support that remain an unmet need for patients and caregivers and make recommendations to the Scientific Advisory Board.
    - Be the voice of the patient and caregiver on GARDIAN, providing feedback on GARDIAN's accessibility, community value and understanding of the registry and its purpose.
    - Empower patients and families to engage with and drive the direction of research and scientific advancement on nGD.
    - Identify how GARDIAN can best provide feedback to the community on its activities and make recommendations to the Strategic Planning & Registry Development.
    - Review the GARDIAN website content and make recommendations for its development.

FINANCE:

The IGA is funded through grants and sponsorship from a number of pharmaceutical companies 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.



In 2021, to support the development of the GARDIAN Registry, the IWGGD and the IGA provided grants to the new company International GARDIAN Ltd (a company wholly owned by IGA) for a period of two years (2021-2023) to support the employment of the Communications and Campaigns Officer and the Projects Officer for 15 hours a week combined. This is reflected in our report this year as a payment from IGA.

Our board of Directors, CEO, volunteers and doctors have donated over 2083 hours of their voluntary time to our activities.

THIS IS EQUAL TO £52,086.

We are extremely grateful for this dedicated support.

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, Freeline, Gain, Lilly, M6P, Pfizer, Sanofi, Takeda.

IN 2023 WE INTEND TO:

- Revise our Strategic Plan for the next five years to ensure that our values, vision and mission remain relevant and that our activities and campaigns are effective, that we are serving our Members and Gaucher patients and caregivers around the world in the best way possible, and that we continue to improve the visibility and knowledge of Gaucher disease. In 2022 we consulted our community and other stakeholders, and in 2023 a working group will create a revised document for 2024 to 2029 based on the feedback.

- Hold a Go With Gaucher symposium for young adult Gaucher patients, from IGA Member organizations, giving them a unique opportunity to meet their peers and develop their skills.

- Continue our work with the older generation of Gaucher patients to have a better understanding of their clinical and non-medical needs and to detect unmet needs and improve their situation where we can.

- Develop Best Practice switching and monitoring guidelines, in co-operation with the International Working Group on Gaucher Disease (IWGGD), that can be used by patients to take to their clinicians to ensure that if they are made to switch to a new medication this is done in the best and safest way possible for the patient.

- Expand International Gaucher Day, with a theme based around diagnosis which will include a keynote lecture on the theme and a video to be broadcast on the day.

- Issue a statement on non-comparable treatments for Gaucher disease to ensure that all Gaucher patients have access to safe and effective medication.

- Continue to expand our Volunteer and Regional Manager programmes, including educational programmes for training and capacity building.

- Build on our gene therapy educational programme to reach more patients, caregivers and healthcare providers.

- Publish a paper on the self-management of Gaucher disease and continue our Homes Not Hospitals campaign.

- Support our Members and expand the IGA's reach by translating more documents into more languages.

- Expand our efforts to educate and empower our Members through webinars, publications and other means.

- Build on our relationships with the pharmaceutical industry by:
    - working to ensure humanitarian access to treatment for as many patients as possible.
    - raising awareness of clinical trials
    - highlighting unmet needs
    - participating in advisory boards
    - collaborating on projects and joint initiatives for the benefit of the community

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