IGA INTERNATIONAL GAUCHER ALLIANCE

Driven by passion for patients

IMPACT REPORT 2021

GARDIAN

THE GAUCHER NEURONOPATHIC REGISTRY

OWNED AND GOVERNED BY THE GLOBAL PATIENT COMMUNITY

Together we are stronger

INTRODUCTION:

We all went into 2021 not really knowing what to expect, the global COVID pandemic was still raging across the globe and sadly 2021 saw another 12 months of uncertainty, movement restrictions and many people feeling isolated from friends and family.

However, as a community we came together to ensure that patients felt supported, were able to access treatment and our doctors and healthcare professionals worked tirelessly to ensure that patients were able to contact them, often using online platforms, which have become such a big part of all our lives. The pandemic meant that all meetings were hosted online, and the lack of physical contact has been hard for everyone. Mental health has been high on everyone's agenda and by supporting our community with regular webinars, we hope that this was of some comfort.

2021 saw two new members of staff join the IGA team to support our communications strategy, Harry, and Wendy for the regional manager programme and GARDIAN Champion programme, both for the IGA and for the new global GD2 and GD3 registry GARDIAN which we will launch in early 2022.

With a commitment to our members to advocate for home therapy and building on the success of our 2020 IGD campaign '***Home not Hospitals, improving patients' quality of life by campaigning for home therapy***', we held an online members meeting to share experiences and raise awareness of the work of the IGA in collaboration with the EWGGD on developing global guidelines for home therapy.

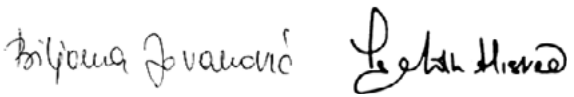
We forged ahead with our gene therapy project, increasing community understanding and education on Gene Therapy to improve recruitment for clinical trials, and will take this through to 2022 where we will use the information from our focus groups and global survey to inform the development of educational materials on gene therapy for our community.

October saw another successful IGD with our focus on improving diagnosis '***Early diagnosis, better lives***'. We are extremely grateful to our community who shared their stories to raise awareness of the diagnostic odyssey that remains an issue for patients and families and can have a detrimental effect both physically with possible irreversible damage and mentally knowing something is wrong but not what it is.

The IGA has a very close relationship with the European Working Group on Gaucher Disease (EWGGD) and this was strengthened further with the development of five working groups, made up of clinicians and patient advocates, for the development of management guidelines for GD1 and GD3, as well as an series of ethical workshops on topics such as clinical trials in countries where there is no access to treatment, newborn screening, and gene therapy.

2021 ended with a change in the Chairmanship of the IGA with Vesna Aleksovska standing down as chair to take up a position on the IGA staff team as Projects Officer and Vice Chair Biljana Jovanovic becoming the new Chair.

The IGA's logo driven by passion for patients, is demonstrated with a huge increase in the number of volunteers that have joined us in 2021 to enable us to have a much wider reach and meet the needs of our diverse community across the globe.



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 **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.

PATIENTS

IGA INTERNATIONAL GAUCHER ALLIANCE
Driven by passion for patients

DOCTORS & HEALTHCARE WORKERS

Our MISSION

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

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

Our VALUES

We are:

- Visionary
- Patient focused
- Passionate
- Trustworthy
- Collaborative

IN 2021 WE MADE THE DIFFERENCE BY:

- Hiring a Communications and Campaigns Officer to help us to spread our messages more widely and to raise the profile of the IGA.
- Hiring a Projects Officer to support the recruitment and retention campaign of the nGD community to sign up to the global registry GARDIAN as well as the Regional Manager and Volunteer programmes.
- Translating the basic Gaucher disease information booklet and "*What being a member of the IGA can do for you*" document into seven languages: Spanish, Chinese, Arabic, Hebrew, French, Russian, Portuguese. They are available to download from the IGA website.
- Raising awareness about Gaucher disease through International Gaucher Day – on 1 October we were able to reach over **4000** people on Twitter; **4000** people on Instagram and **12,000** people on Facebook.
- Increasing community understanding and education on Gene Therapy to improve recruitment for clinical trials. The IGA engaged UrbanKind Institute to professionally facilitate a number of focus groups limited to 4-7 individuals to best encourage participation. One of the groups included translators to ensure non-English respondents could participate.

Providing the following support for members:

» Ukraine – procurement process of medicines for GD, patient and doctor driven rather than budget driven decisions

» Peru – patient rights to choose best medicines for patients

» Mongolia – access to oral therapy due to challenges with importation of ERT

» South Africa – supporting access to information to support patient decision making

» Morocco – re instatement of government funding for treatment for GD patients

» Supporting patients in non-member countries including Algeria, Rwanda and Sudan

IGA volunteers made a difference by:

- Starting the IGA GARDIAN Champions Programme, an initiative designed specifically to support the growth of the GARDIAN registry by encouraging patients and caregivers to sign up. GARDIAN Champions are GD3 patients or have a close connection to GD2 or GD3 (e.g. a family member, friend, caregiver, or medical professional.) The success of GARDIAN will depend on patients and caregivers' support in signing up to the GARDIAN registry, and Champions will play a vital role in encouraging and supporting people through the process. At the end of 2021 we have successfully recruited champions from the USA, the UK, Israel, Italy, Denmark, Argentina, Greece and Egypt.
- At the end of 2021, **41** volunteers from **34** countries were involved in over 11 projects, accounting for **1373** volunteer hours. Most of the volunteers (around 70%) were recruited through advertisements on social media and some of them were directly invited to join in projects and activities. Volunteer activity included the Go With Gaucher project, the home therapy project, translations, the Regional Manager Programme,

the older generation project, the co-morbidities project, the global patient profile, GD1 guidelines, membership meetings and GARDIAN Champions.

- Reviewing and reforming the IGA Regional Manager (RM) Programme, beginning the process of recruiting new RMs. 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.

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

Publications:

- A charitable access program for patients with lysosomal storage disorders in underserved communities worldwide. Tanya Colin-Histed as a member of the Takeda Medical Expert Committee was an author.

Advisory Boards:

- Avrobio: GD3 educational session for senior leadership team
- Sanofi Genzyme GD3 advisory Board
- GOS: Gaucher outcome survey for Takeda
- ICGG: International Collaborative Gaucher Group (ICGG) Gaucher Disease Registry

Conference/Event speaker:

- Takeda's Charitable access medical tutorial: The role of the IGA in charitable access countries
- Rare2021: GARDIAN
- Rare Disease Day 2021: ethical and social justice-related issues in rare diseases *'when every step is a fight for equity: the value of patient advocacy'* University of Michigan, USA
- FDA Listening Session Neuropathic Gaucher Disease with Gaucher Community Alliance (GCA) in the US
- Pakistan Gaucher preceptorship at Lahore's children's hospital in Pakistan: Unmet needs of patients in developing countries and access to treatment opportunities.
- Access to medicines in Africa: What is important to patient? Diagnosis

Collaborating:

- Worked with the IWGGD on a collaborative approach to developing international clinical patient-centric guidelines for Gaucher disease.
- Takeda Blueprint meeting in March 2021, The importance of patients/caregivers and the national patient group in building capacity, involving CEO and members from Botswana and Pakistan
- European Haematology Association (EHA) : presentation at their UK educational Haematology Gaucher seminar on GD and COVID -19
- 11 pharmaceutical companies with an interest in GD, mostly education, clinical trial design, awareness, patient literature, introductions to other members.
- PQMD: Global Access to Rare Diseases (GARD) project
- Representation at the Global Genes Patient Advocacy Summit virtual conference

Training

- Clinigen's early access fellowship programme

KEY ACTIVITIES:

HUMANITARIAN AID

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.

Direct requests received for support to access treatment to IGA: **11** countries including Turkey, Sudan, Saudi Arabia, Rwanda, Peru, Pakistan, Nicaragua, Morocco, Kenya, Jordan, and India.

- Three deaths before treatment could be found
- 2 cases have been approved, awaiting treatment to begin in 2022
- 1 adult case, outstanding, challenge to access charitable treatment for adults
- 2 patients moved back to ICAP after government funding ceased to be available
- 3 treated
- 1 awaiting confirmation of diagnosis

At the end of 2021 we have 10 patients' cases on our database that we have not been able to support..

The challenges our community continue to face:

- Little opportunity for adults to access charitable programmes
- Treatments being registered in countries by the pharmaceutical industry, but no government reimbursement programmes in place
- Increased education of clinicians and diagnosis, therefore an increase in the numbers of patients being diagnosed, however no treatment reimbursement in too many countries, relying on charitable access programmes for treatment but the programmes have limited capacity
- Challenge to diagnostics for patient (they have to pay) and for doctors where there are no facilities in their countries

The IGA works collaboratively with the three pharmaceutical companies that provide charitable treatment to Gaucher patients, these are:

- Takeda Charitable access programme: through the Medical Expert Committee (MEC) programme (the IGA is a member of the MEC) in 2021 a total of **17** patients were approved from Sudan, Pakistan, Kenya, Tanzania, Tunisia, Indonesia, and Palestine
- Pfizercares: one new referral approved for funding, with a further 3 in the pipeline
- Sanofi Genzyme's ICAP: 3 patients approved for treatment. Please note that this figure only includes those patients referred to Sanofi Genzyme 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

INTERNATIONAL GAUCHER DAY

Our 2021 campaign focussed on the challenges Gaucher patients are facing in getting a timely diagnosis.

We shared the stories of patients from around the world, both is social media posts and in videos. We also shared the views of clinicians and other working in the field.

We made a total of **36** Facebook posts and another on **36** Instagram. We sent **26** Tweets. and made **8** LinkedIn posts.

We also shared five videos in different languages.

As well as our own posts, members and partners also posted, with some translating the information into various languages.

INTERNATIONAL GAUCHER DAY 1 OCTOBER
International Gaucher Alliance
Rare but not alone

Social Media posts during International Gaucher Day 2021

GARDIAN:

GARDIAN
THE GAUCHER NEURONOPATHIC REGISTRY
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Gaucher Registry for Development, Innovation and Analysis of Neuronopathic disease (GARDIAN)

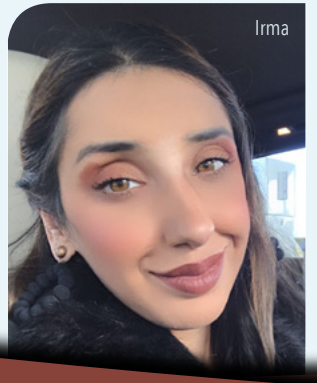
In 2020 the IGA set up a company called International GARDIAN limited (IGL). IGL wholly owned by the IGA, therefore the global gaucher community will own and govern a global patient registry for Types 2 and 3 Gaucher disease.

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.

2021 saw a huge leap forward to launching the registry:

- We undertook cognitive interviews with the patient and caregiver community in the 8 languages that GARDIAN will be available in to ensure that the translations of words are relevant and that the way the questions are asked are understandable
- We appointed a communication and campaigns officer (5 hours a week) and a project officer (10 hours a week) to help support the development of GARDIAN
- We held a listening session with the Food and Drugs Administration (FDA) at which 38 persons from the sub real world evidence (RWE) committee attended the 1 hour session to learn more about GD types 2 and 3 and the registry
- We instigated communications with European Medicines Agency (EMA) through their Innovative taskforce (ITF) process to identify scientific, legal and regulatory issues relating to GARDIAN
- We developed our GARDIAN champions programme, using GD3 patients, caregivers, and clinicians, we will engage and recruit the patient community to sign up to GARDIAN
- Working with 67 Health, we developed a GARDIAN toolbox of key messages for patients, caregivers, and the clinical community to promote and understand the value of GARDIAN
- We worked on the consent process for GARDIAN, including a video that will have subtitles for translations, we developed a baseline questionnaire to collect demographic and clinical data and the verification process to ensure only eligible GD2 or GD3 patients and caregivers are accepted into the registry
- In late October, the Institutional Review Board (IRB) was successfully approved for GARDIAN, and work began immediately on building the registry platform. The IRB is a committee established to review and approve applications for research projects involving human subjects. The primary purpose of the IRB is to protect the rights and welfare of the human subjects
- Initiated a paper for publication titled: A Global Neuronopathic Gaucher Disease Registry: A Patient-Led Initiative, with the Target Journal: The Patient - Patient-Centered Outcomes Research

Hi my name is Irma and I am an GD type 3 patient. By being a Gardian Champion I will be helping and supporting patients to register. This is very important to me as I believe it's very important to rise awareness about nGD and gather patient centric data to improve lives. Raising awareness will help clinicians and patients as nGD is a rare disease and many still have no knowledge about it.



EDUCATIONAL WEBINARS:

The IGA continues to support the Gaucher community to increase their capacities through education and providing member organisations, patients, families and healthcare professionals with information about all aspects of Gaucher disease. In the regular communication with members and through survey, topics for webinars in 2021 were identified. These meetings were primarily for Gaucher patients and families but were open for all interested, including clinicians and pharma representatives. Speakers were open to questions at the end of each session.

1. Wellness for Gaucher disease

Two speakers talked about how to improve the everyday lives of Gaucher patients

- Dr Seema Kanwal, a Naturopathic doctor and speaker, talked about maximising patients' well-being. Seema has been a practising naturopathic doctor in Vancouver since 2006 and has extensive experience with and is passionate about working with individuals who have rare genetic diseases and suffer many stress-related conditions as a result such as mental health issues, insomnia, and low energy
- Janez Vajevec, an acting coach talked about body and feelings relaxation and demonstrated relaxation and meditation techniques

2. Bone disease

Bone involvement is one of the most prevalent aspects of GD and a major cause of pain, disability, and reduced quality of life. All three types of Gaucher disease have bone disease in common and can have serious complications in those affected. Existing therapies for Gaucher disease (ERT and SRT) are very effective in reversing some of the clinical manifestations of Gaucher disease such as anaemia, thrombocytopenia, etc. but skeletal responses are much slower. Some patients even after many years of treatment, still have bone crisis, bone pain or severe symptoms.

- Dr Patrick Deegan, MD, FRCP Edin, FRCPath from Addenbrooke's Hospital Cambridge University Hospitals NHS Foundation Trust gave an introduction to bone disease
- Dr Pramod Mistry, MD, PhD, FRCP, FAASLD from Yale School of Medicine talked about maintaining bone health through generations of Gaucher disease

3. Mental health

This webinar was open to young Gaucher patients, as a part of the Go with Gaucher project. Dr Stacey J.Feuer, PsyD, MLD, Director of Health Psychology talked with young people about mental health and teach them tips and tools for dealing with different issues.

4. Biomarkers

Biomarkers are objective medical signs used to measure the presence or progress of the disease or the effects of treatment. In lysosomal storage diseases such as Gaucher disease, there are several available biological markers that are useful tools in diagnosis that offer objective information about the situation of the disease and in the follow-up to detect early complications and the stability in the response to treatment. This webinar was about biomarkers in Gaucher disease and Lyso-GB1. Speakers:

- Professor Maria Fuller- Biomarkers in Gaucher disease, BAppSc, MAppSc, PhD, FFSc(RCPA) National Referral Laboratory, Genetics and Molecular Pathology, SA Pathology, Adelaide, South Australia
- Professor Ari Zimran MD LysoGb1 as a key parameter in the management of patients with GD, Associate professor of Medicine (Gaucher Unit) – Shaare Zedek Medical Center

5. Fundraising

Speaker Jonah Halper, MPA is a thought leader on nonprofit fundraising and marketing, specializing in new donor acquisition and engaging new leaders in 21st-century philanthropy. Through the lens of personal relationships, Jonah guided

EDUCATIONAL WEBINARS:

attendees through 21st-century donor cultivation and solicitation, and how it relates to our organizations in a world where donors have so many philanthropic options - and much less attention span.

6. nGD – the spectrum, the experience and the therapies

The focus of this webinar was on nGD. Three fantastic speakers gave lectures on the following topics:

- The spectrum of nGD and what we know. - Raphael Schiffmann, MD, MHSc, FAAN is an expert on neurometabolic diseases
- The experience of Ambroxol in nGD, what do we know - Ari Zimran, the Founder and the Director of the Gaucher Unit at Shaare Zedek Medical Center in Jerusalem, Israel from 1990 to 2018, and is currently a senior physician at the Unit
- New potential therapies for nGD - Derralynn Hughes, Professor of Experimental Haematology at the University College London, Clinical Director of Research and Innovation at the Royal Free London NHS Foundation Trust, and Co-Clinical Director of the NCL cancer Alliance

7. Gene therapy

A members-only meeting on the topic of Gene therapy for Gaucher disease. The objective of the webinar was to educate IGA members on the different approaches and discuss some of the challenges of Gene Therapy. The aim was to educate IGA members so that they can make informed decisions should they wish to consider gene therapy for themselves or their child (ren). The main presentation was given by Professor Atul Mehta, and he was available for Q&A afterwards.

Webinars that are for the wider community are uploaded to the IGA YouTube channel.

INDIVIDUAL CONTRIBUTIONS:

Living with Gaucher is balancing life and Gaucher. Enjoying life, I returned to Gaucher to serve others less fortunate than me. Covid provided me with a lot of extra free time. I enjoy working together now with the other Gaucherians while doing what I like best: researching and writing. Gaucher can be fun!

PAUL

I decided to volunteer for the position of IGA Africa Regional Manager because I felt that Gaucher is under reported in Africa and being one of the few rare diseases with some treatment options, awareness would help both clinicians and patients' families obtain an early diagnosis which would hopefully lead to early interventions. I was also interested in working with an international rare disease organisation to learn new skills in the field. I look forward to being a part of the IGA team.

ROSELYN

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.

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 recieving no further funding, to continue to serve our global community and implement a strategy to address how to take the IGA forward.

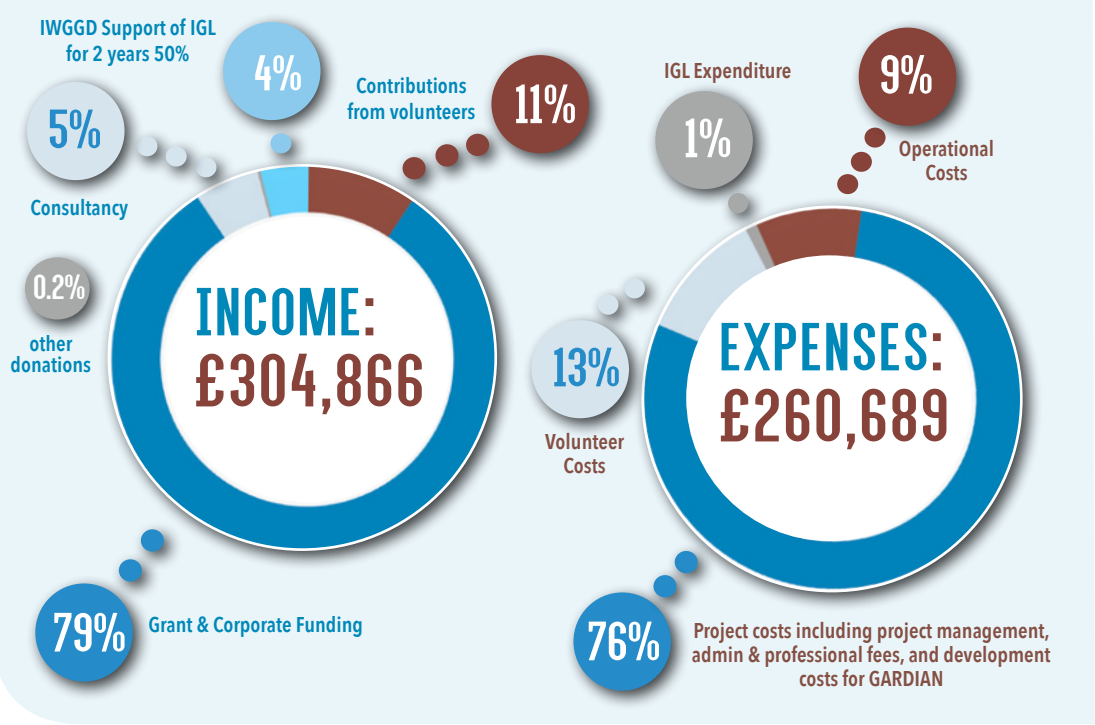
The work of the IGA Board of Directors is supported by very dedicated and passionate staff: Chief Executive Officer, Operations Officer, Communications and Campaigns Officer and Projects Officer.

Due to the Covid-19 Pandemic we unfortunately could not undertake every project we had planned for 2020/2021 so we will carry over these projects to the following year.

Our board of Directors, CEO, volunteers and doctors have donated over **1373** *hours of their voluntary time to our activities.*

IT IS EQUAL TO £34,345

We are extremely grateful for this dedicated support.



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 2 years to support the employment of the communications and campaigns officer and the project officer for 15 hours a week. This is reflected in our report as a grant from the IWGGD and payment from IGA.

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!

Avrobio, Eurordis, Freeline, Gain, M6P, Orphazyme, Oxyrane, Pfizer, Prevail, Sanofi Genzyme, Takeda.

IN 2022 WE INTEND TO:

- The IGA will continue with educational sessions for the Gaucher community on topics that are identified as of interest to patients and families. As the IGA is a global organisation, educational materials will be recorded and available online on social media platforms, for the community to watch when they can. It is planned to record lectures from main researchers but also sessions with IGA members sharing their experience and good practice
- Translations of main IGA documents will be continued, for 2022 it is planned to translate the Best practice brochure, IGA Code of practice, Strategic plan and Impact report to at least two languages
- Bring all our members together on the 7th and the 8th of May in Leiden, The Netherlands for our biennial members meeting that will be face to face and also offer online access
- Launch the GARDIAN registry to increase knowledge and awareness and continue to improve the lives of nGD patients and their care givers
- Raise awareness of Gene Therapy and understand the patient's view
- Implement several guidelines, with IWGGD, to help GD patients and physicians (GD1 and GD3 guidelines)
- Strengthen our relationship with pharma industry (regular meetings)
- Take an active part in evaluating safety and efficacy of the Gaucher drugs (registries)
- Continue the work in Africa with the Access to Medicine project, that began in 2021 by providing 100 free diagnostic tests for patients with RD in collaboration with FYMCA, North West University, Potchefstroom and Rare Disease South Africa
- Undertake research into the challenges of our maturing Gaucher generation to empower, share and strengthen both Gaucher patients and the IGA member organizations to enable our older community to live their best life
- Bring together young adult Gaucher patients at a face to face meeting to exchange information and ideas through our long standing ***Go with Gaucher*** programme
- Introduce a mentoring programme, to support those individuals who run patient groups to achieve their own goals by ensuring they have the appropriate skills, knowledge, and confidence
- Expand our regional manager programme into areas of the Middle East

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