Global Collaborative Effort for Gaucher Disease:
A Step Towards Improving Healthcare

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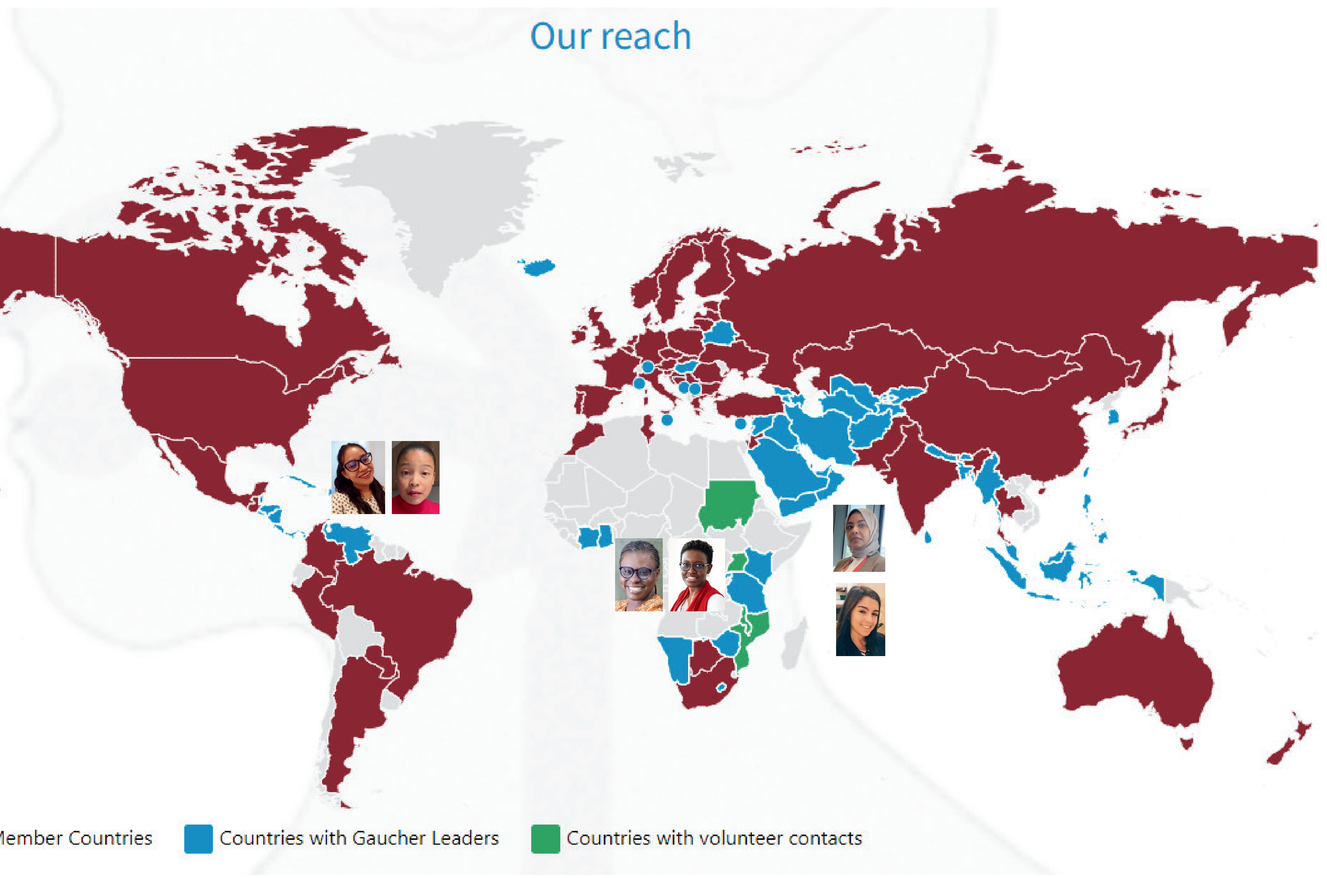
Collaborators

International Gaucher Alliance (IGA), A Rare Cause (ARC), International Working Group on Gaucher Disease (IWGGD), SymetryML, Rare Diseases Forum Sri Lanka (SLCP), Rare Disease Society Ethiopia, Metabolic Support UK



The Global Gaucher Connect Programme

The Global Gaucher Connect Programme collaborates with healthcare professionals and organizations to address challenges faced by patients and their families.

By working with volunteer advocates, the IGA has been able to meet the growing and diverse needs of our global community whilst recognising the challenges of limited resources and the importance of understanding the language and culture of a region/country is imperative to build sustainable communities.

Challenges faced by GD patients in Sri Lanka and Ethiopia

- Limited access to diagnosis
- Low awareness among healthcare providers
- Restricted access to treatment

Opportunities for improvement in Sri Lanka and Ethiopia

- Increasing Awareness: Educating healthcare professionals and the public about GD
- Improving Access to Treatment: Advocating for supportive government policies
- Building Capacity: Strengthening local healthcare systems and developing patient organisations
- Developing Clinical Networks: Connecting healthcare professionals with supportive services

Sri Lanka: A Path Towards Better Care

Dr Duminda Samarasinghe, President SLCP

- Establishing a national rare disease registry (including GD) with the support of SymetryML by using federated AI technology
- Education and awareness initiatives with the Rare Disease Forum
- Using registry data to:
  - Understand GD prevalence
  - Identify treatment needs
  - Improve care through education and awareness
  - Advocate for policy changes
- Establishing a newborn screening programme with the help of Metabolic Support UK

Visit the Metabolic Support UK website

Visit the RDF website

Visit the SymetryML website

Overcoming barriers: Bridging the gap between global and local needs in Lysosomal Storage Diseases with focus on Gaucher Disease

The Rare Disease Forum of the Sri Lanka College of Paediatricians and the International Gaucher Alliance (IGA) hosted a hybrid session on Lysosomal Storage Disorders (LSD), focusing on Gaucher Disease. The aim was to raise awareness about diagnosing and managing LSD in children and providing charitable access to enzyme replacement therapy. Held at Lady Ridgeway Hospital, Sri Lanka's top children's hospital, the webinar had over 100 in-person attendees and around 50 online participants.

Developing a registry in Sri Lanka

The Registry Application is a tool for healthcare professionals managing rare disease patients. It uses the FHIR format to streamline data documentation and management, enabling efficient tracking of patient encounters, observations, lab results, and diagnostics. Physicians can link their practice to multiple organizations, and its reporting dashboard offers insights into disease burden and prevalence. The registry leverages SymetryML's Federated AI platform for secure data collaboration and advanced analytics, enabling global partnerships without moving data.

Building Hope in Ethiopia

Education and awareness initiatives (webinars/workshops) with the support of A Rare Cause (ARC)

- Promoting early diagnosis with DBS cards with the support of ARC
- Advocating for access to medications
- Building partnerships with local experts and supporting development of patient group in cooperation with the doctors' organisation in Ethiopia
- Building a network in Africa and promoting sharing experiences, technologies, and knowledge through the Africa Roadmap

Visit A Rare Cause website

Online educational seminar

On May 30, the IGA and A Rare Cause organized an online seminar with the Ethiopian Endocrine and Metabolism Society on Gaucher and other inherited metabolic diseases. The event had 148 engaged participants, enhancing understanding of diagnosis and treatment challenges in Ethiopia and Africa.

Ethiopian doctors emphasized the importance of diagnosing Gaucher Disease (GD) using DBS (Dried Blood Spot) cards through A Rare Cause. Currently, Lancet Laboratories and DHL provide some logistical support, but expansion and more information are needed. Training additional local personnel is a long-term goal.

The IGA's efforts in Ethiopia and Sri Lanka are significantly impacting individuals with Gaucher disease by proactively and reactively addressing challenges and fostering a sustainable, inclusive and supportive healthcare framework for rare diseases, working together with reliable partners.

About 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 and has built its reputation through listening to and delivering outcomes that have impacted on patients and their carers' lives. The IGA is an independent charity with a clear code of practice for working with industry. In 2024, the IGA celebrated its 30th anniversary.

Visit the IGA website

Read about GD on the IWGGD site