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

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Background

- Gaucher Disease (GD) is a rare inherited metabolic disorder.
- Type 2 and Type 3 are neuronopathic and often result in infant death or progressive neurological deterioration.
- Current drug therapies do not cross the blood brain barrier and thus do not treat neuronopathic GD (nGD).
- GARDIAN data will generate real-world evidence on the natural history and impact of disease and will inform clinical trial design and healthcare decision making.

Objectives

The objective is to describe the development of a patient registry specific to nGD. The registry aims to:

- Improve our understanding of the demographic, and disease-related characteristics of patients with nGD.
- Improve our understanding of the self-reported burden over time, including changes in symptoms and health-related quality of life (HRQoL) for patients with nGD.
- Develop and validate disease-specific outcome measures for assessing HRQoL over the natural course of Gaucher Disease.
- Report results of GARDIAN to stakeholders, including the nGD communities.

Methods

- The International Gaucher Alliance (IGA), patients, caregivers, clinicians, and researchers partnered to develop a web-based platform for patients with nGD and their caregivers.
- Baseline and follow-up questionnaires were designed to capture data relevant to patients, including newly developed nGD-specific Patient Reported Outcome (nGD-PRO) and Observer Reported Outcome (nGD-ObsRO) instruments to be validated within the registry.
- Qualitative interviews were conducted to ensure the use of terminology relevant to patients.
- Diagnosis confirmation processes were informed by clinicians.

Registry flow

GLOBAL RECRUITMENT:

- Patients with nGD and caregivers are approached globally by IGA local member associations/bodies which share information about the registry using email, social media, and other electronic platforms.

ONLINE REGISTRATION:

- Patients and caregivers register with automatic account verification and provide consent to complete registration to the online platform accessed using a computer, smartphone, or tablet.
- Upon consent, the participants are prompted to provide a proof of diagnosis, such as enzyme / genetic results or a physician's report, which is reviewed by a dedicated medical team.

DATA COLLECTION:

- Participants who qualify as eligible are enrolled and able to complete a questionnaire at baseline and again 10 to 14 days later.
- Participants will be asked to complete follow-up surveys every 6 months, up to 3 years.

Stakeholder engagement

Clinical experts
- Advise on the registry objectives, data acquisition and data analyses and provide guidance on the registry conduct
- Review and evaluate registry-related materials

IGA / IGL
- Holds legal responsibility and makes strategic decisions regarding GARDIAN.
- Advertising and promoting the registry recruitment

nGD patients and caregivers
- Qualitative interviews with patients and caregivers using the registry-related questionnaire to ensure the use of relevant terminology

Researchers
- Ensure the registry management
- Identified and designed to capture the relevant health burdens and concerns that were experienced by nGD patients and caregivers

Industry
- Donations and grants to the registry

Data collected

Key clinical characteristics

- **Genetic testing** in the glucocerebrosidase (GBA) gene (date & results)
- **Neurological and non-neurological symptoms** at diagnosis, at baseline and longitudinal follow-up

Patient characteristics

- Demographics (gender, age, region, ethnicity, country of origin)
- Social and economic indicators (level of education, occupation…)

Disease characteristics

- Medical history (age at diagnosis, symptoms at diagnosis, age at onset of puberty)
- Type of nGD (Type 2/Type 3)
- Family history of GD
- Family history of Parkinson, other comorbidities
- GD characteristics/current symptoms

GD treatment /surgeries, procedures

Aids/home adaptations

- Health and social care services used

Patient Reported Outcome (PRO) and Observer Reported Outcome (ObsRO) measures

- PRO and ObsRO specific to neuronopathic Gaucher Disease (nGD-PRO/ nGD-ObsRO) (new measures that were developed and are being validated as part of GARDIAN): Pain, Fatigue, Symptoms, Emotional distress, Functionality, Disability, Treatment concerns, Experience of care, Quality of life (patients and caregivers)
- PedsQL (Pediatric Quality of Life): Physical functioning, Emotional functioning, Social functioning, School functioning
- PGI-S (Patient Global Impression of Severity): Severity
- PHQ-9 (Patient Health Questionnaire): Depression
- GAD-7 (General Anxiety Disorder): Anxiety

Inclusion criteria

Patient Criteria

- Confirmed nGD diagnosis.
- Regular access to the internet and functioning email account.
- Patient (or legal representative) provides online informed consent to participate.
- Participants who can read and understand the study specificities provide online consent to participate in the registry.

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Caregiver Criteria

- Age 18 years or older.
- Primary or co-primary caregiver of a patient with a confirmed nGD diagnosis.
- Legal authorized representative of the patient with a confirmed nGD diagnosis.
- Regular access to the internet and functioning email account.
- Provides an online-informed consent to participate in the registry.

Registry languages

- English
- French
- German
- Spanish
- Arabic
- Japanese
- Chinese

Acknowledgments

This registry is conducted by Cerner Enviza on behalf of the International Gaucher Alliance (IGA) and multiple stakeholders.

Results

- The patient-initiated Gaucher Registry for Development Innovation and Analysis of Neuronopathic Disease (GARDIAN) is a global prospective patient registry with stakeholder-defined objectives.
- Data are collected at baseline and every 6 months for 3 years.
- Data include patient-reported enzyme/genetic results, patient characteristics, symptoms, medical history, treatment, comorbidities and patient- / caregiver-reported outcomes.
- GARDIAN was approved by institutional review boards.
- Patient enrollment began in April 2022.

Conclusions

- The systematic and standardized collection of real-world data will provide a research platform for improving disease understanding, advancing disease management, designing treatments, and improving patient outcomes.
- Patient engagement in the development of GARDIAN optimizes its value as a real-world data source to inform healthcare decisions and address the unmet needs of patients.

Scan for GARDIAN website

Scan for GARDIAN fact sheet