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.

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.

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



engagement

Stakeholder

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



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

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 diseaserelated 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.

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

Methods

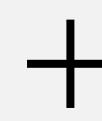
- The International Gaucher Alliance (IGA), patients, caregivers, clinicians, and researchers partnered to develop a web-based platform for patients with nGD and their caregivers.
- Baseline and follow-up questionnaires were designed to capture data relevant to patients, including newly developed nGD-specific 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.

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.



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
- Arabic
- French German
- Japanese Chinese
- Spanish

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.















