Characteristics of patients with Type 3 Gaucher Disease (GD3) and use of medical devices and supportive services: Baseline results from the Gaucher Registry for Development, Innovation and Analysis of Neuronopathic Disease **(GARDIAN)**

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Background

- Gaucher Disease (GD) is a rare inherited metabolic disorder. Type 2 (GD2) and Type 3 (GD3) are neuronopathic and cause infant death or progressive neurological deterioration.
- Current drug therapies do not cross the blood brain barrier and do not treat neuronopathic GD (nGD).
- GARDIAN is a longitudinal, global patient registry developed by the International Gaucher Alliance (IGA), patients, caregivers, clinicians, and researchers.
- Patient- and caregiver-reported data are collected on enzyme/genetic results, patient characteristics, neurological and non-neurological symptoms, medical history, treatment, and comorbidities.
- Patient- and caregiver-reported outcomes include the Pediatric Quality of Life (PedsQL), Patient Global Impression of Severity (PGI-S), General Anxiety Disorder (GAD-7), Patient Health Questionnaire (PHQ-9) and an nGD-PRO and nGD-ObsRO to be validated within the registry.
- The data presented here focus on patient characteristics and the use of medical devices and supportive services among patients with GD3.

Objectives

To describe the characteristics of GD3 patients in relation to education, work, and use of medical devices and services as reported at baseline (registry entry).

Methods

- The registry inclusion criteria are displayed below. Eligible patients or their caregivers registered and provided consent to participate.
- A proof of GD3 diagnosis (e.g., enzyme/genetic results or a physician's report) was uploaded by patients/caregivers and confirmed by a dedicated medical team.
- Patients or their caregivers completed an online baseline questionnaire which captured data on patient characteristics, educational level, employment status, living situation, and the use of medical devices and support services.

Inclusion criteria

Patients

- 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.

Parents/caregivers

- 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.

Results

- Baseline data were collected on 25 GD3 patients through early April 2024 as reported by patients (n=10, 40.0%) and parents/caregivers (n=15, 60.0%) (Figure 1).
- The geographical representation of patients is shown in Figure 2, with patients residing in Asia (40%), Europe (40%) and North America (20%).
- Sociodemographic characteristics (including age, gender, ethnicity and living situation) are displayed in Table 1.

Figure 1: Profile of respondents who provided baseline data in GARDIAN

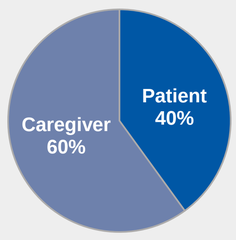


Figure 2: Geographical distribution of patients by continent

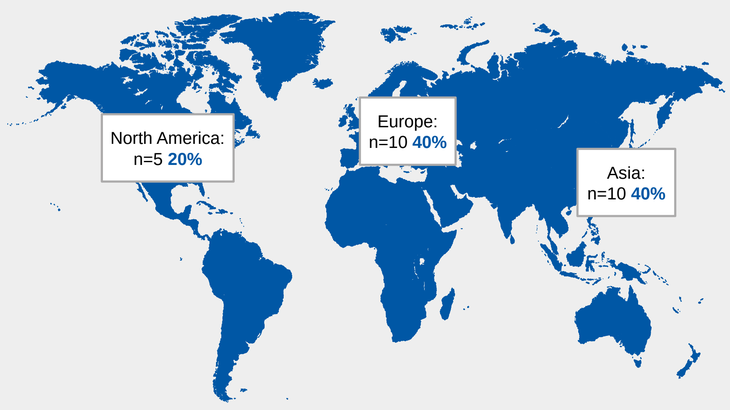


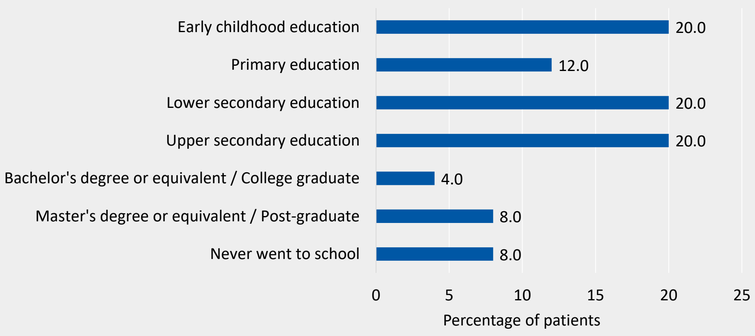
Table 1: Sociodemographic characteristics of GD3 patients

		GD3 patients (N=25)
Age in years, mean (SD)*	At baseline (registry entry)	15.1 (8.4)
Gender, n (%)	Male	13 (52.0)
	Female	12 (48.0)
Ethnicity, n (%)**	White	8 (40.0)
	Asian	4 (20.0)
	Mediterranean	2 (10.0)
	Mediterranean/Arab	3 (15.0)
	Black/African American/Caribbean	1 (5.0)
	Middle Eastern	1 (5.0)
	White/Black/African American/Caribbean	1 (5.0)
Living situation	With parents/siblings	24 (96.0)
	With spouse/partner	1 (4.0)

*Results based on 24 patients due to missing data for one patient.
**Results based on 20 patients (due to local regulations, the question on ethnicity was not be asked in all countries).

The highest level of education obtained by patients is shown in Figure 3. A total of 12% of GD3 patients had obtained a university-level degree. Two patients (aged 5 years and 13 years) had not attended school.

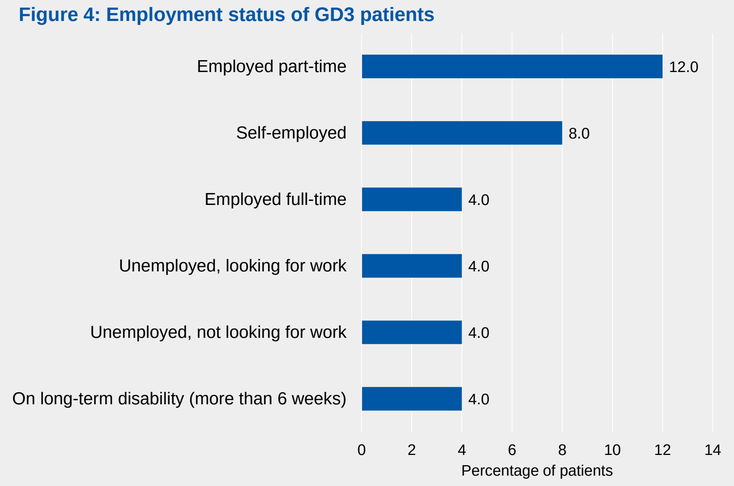
Figure 3: Highest level of education obtained by GD3 patients



Results (cont'd)

- A total of 24% of GD3 patients overall were working (either full-time, part-time or as self-employed). Other patients were unemployed or on long-term disability (Figure 4).
- Employment status was not relevant for 12 patients (48%) who were still in full-time education and 3 patients (12.0%) who were not yet enrolled in school.
- When the 15 patients who were still studying full-time or who were not yet enrolled in school were excluded from the analysis, the percentage of patients working increased to 60%.

Figure 4: Employment status of GD3 patients



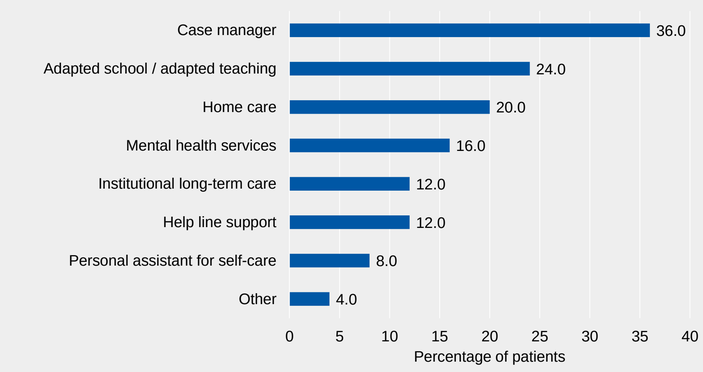
Patients used a range of medical devices, including glasses (n=8; 32%), feet orthoses (n=4; 16%), and wheelchairs (n=4; 16%) (Table 2).

Table 2: Medical devices used by GD3 patients

		GD3 patients (N=25)
Medical devices, n (%)	Glasses	8 (32.0)
	Feet orthosis	4 (16.0)
	Wheelchair	4 (16.0)
	Back brace	3 (12.0)
	Hearing aids	2 (8.0)
	Walk-in shower	2 (8.0)
	Bath lift	1 (4.0)
	Ramps at home	1 (4.0)

- Support services used by patients are displayed in Figure 5. More than one-third of patients (n=9; 36%) had a case manager to assist with paperwork involving health-related services, and almost one-quarter of patients overall (n=6; 24%) had adaptations at school (e.g., modified physical education, extra support at school).
- One patient used 'other' services, which comprised comprehensive care from a multidisciplinary team.

Figure 5: Support services used by GD3 patients



Abbreviations

GD2: Gaucher Disease Type 2
GD3: Gaucher Disease Type 3
IGA: International Gaucher Alliance
nGD: Neuronopathic Gaucher Disease
nGD-ObsRO: Neuronopathic Gaucher Disease-specific Observer Reported Outcomes
nGD-PRO: Neuronopathic Gaucher Disease-specific Patient Reported Outcomes

Conclusions

- GARDIAN generates valuable longitudinal, geographically diverse real-world evidence on nGD patient- and disease-related characteristics.
- The majority of patients (96%) were living with their parents/siblings.
- At registry entry, 40% of patients had achieved a lower secondary or upper secondary education and 12% had completed university.
- Amongst the total registry population, 24% of patients were in employment; this increased to 60% when patients who were still in full-time education or not yet enrolled in school were excluded from the analysis.
- Use of medical devices was common among patients with GD3, with almost one-third of patients wearing glasses.
- A multitude of support services were used by patients and their parents/guardians, including case managers to facilitate access to health-related services, and adaptations in the school setting for patients with GD3.
- The collection of data in a systematic and standardized manner provides a research platform for improving disease understanding, informing clinical trial design, developing safer treatments, advancing disease management, and improving patient outcomes.

Disclosures

Suzanne Reed, Joseph Milce, Perrine Le Calvé, Amina Omri, Amal Sadou, and Bastien Vincent are employees of Oracle Life Sciences, France (a company that received research funds from International GARDIAN Limited), Tanya Collin-Histed is an employee of International Gaucher Alliance.

Acknowledgments

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