

GARDIAN

THE NEURONOPATHIC GAUCHER REGISTRY

OWNED AND GOVERNED BY THE GLOBAL PATIENT COMMUNITY

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Results dashboard for the GARDIAN registry

GARDIAN : Gaucher Registry for Development Innovation and Analysis of Neuronopathic

GARDIAN is a global, longitudinal, prospective, observational registry specific for all patients with **Gaucher Disease Type 2 and 3 (GD2 and 3)**. The purpose of this registry is to better understand Neuronopathic Gaucher Disease (nGD) – its impact on both patients and caregivers and the natural history of the disease, to improve disease management, and support patients and their families. **GARDIAN is owned by the International Gaucher Alliance (IGA)**.



OBJECTIVES

- Improve our understanding of the **demographic, and disease-related characteristics** of patients with GD2 and GD3
- Improve our understanding of the **burden** over time, including changes in symptoms and quality of life for patients
- **Develop and validate disease-specific outcome measures** for assessing quality of life over the natural course of GD2 and GD3 .



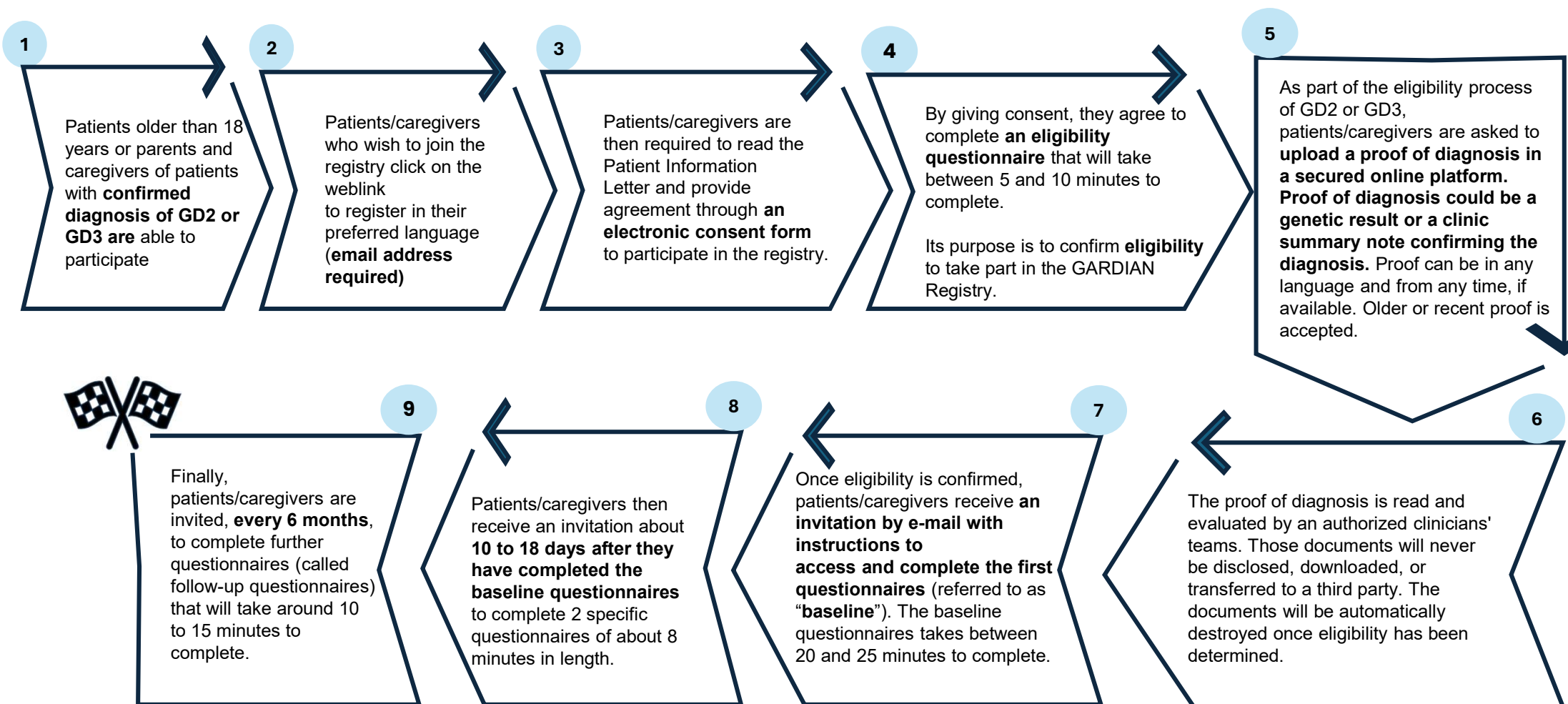
METHODOLOGY

- Patients with GD2 and GD3 and caregivers are recruited through the IGA network.
- Patients complete a baseline questionnaire and then complete follow-up questionnaires every 6 months.
- Data are self-reported by patients/caregivers online

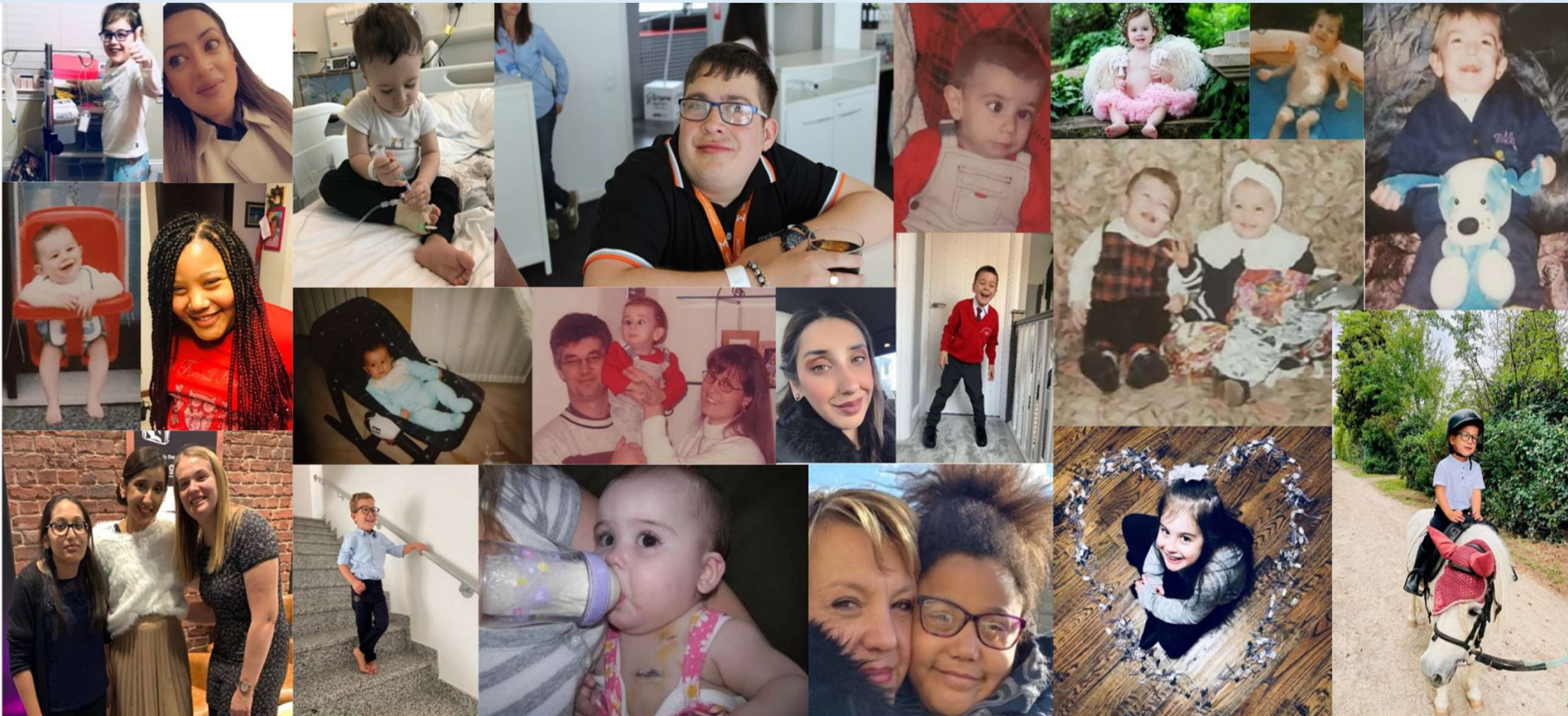
LANGUAGES

Worldwide (7 languages)
English, French, Spanish, German,
Arabic, Japanese and Chinese

Overview of the recruitment and data collection process

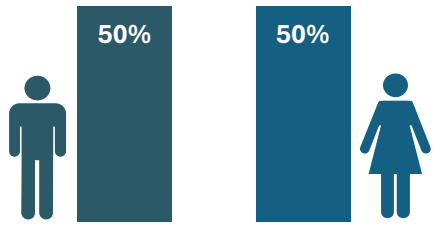


1.1 Profile of respondents

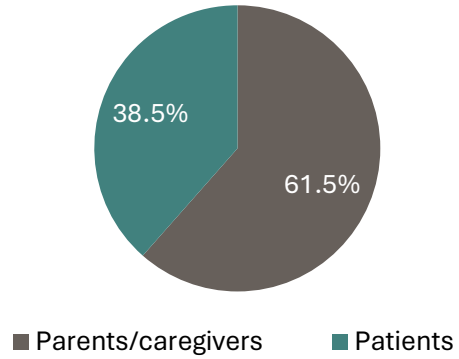


Profile of respondents - Overall

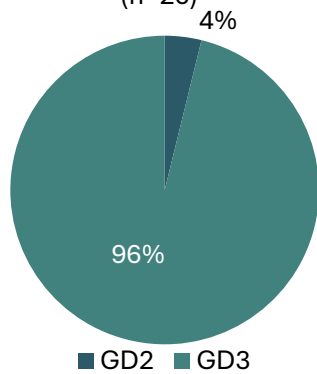
Gender
(n=26)



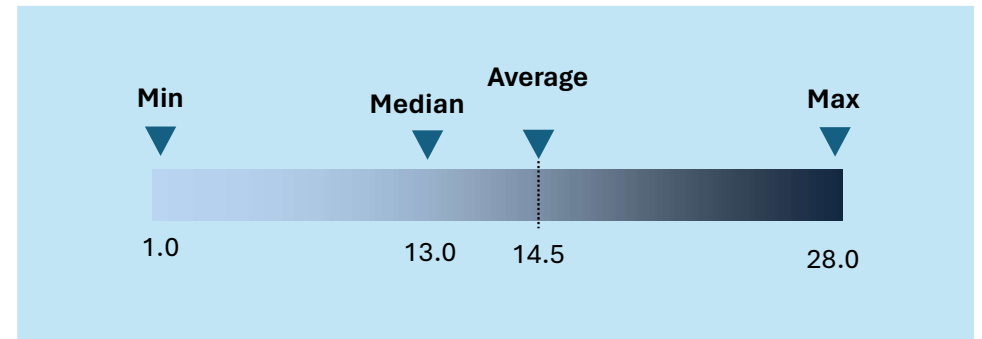
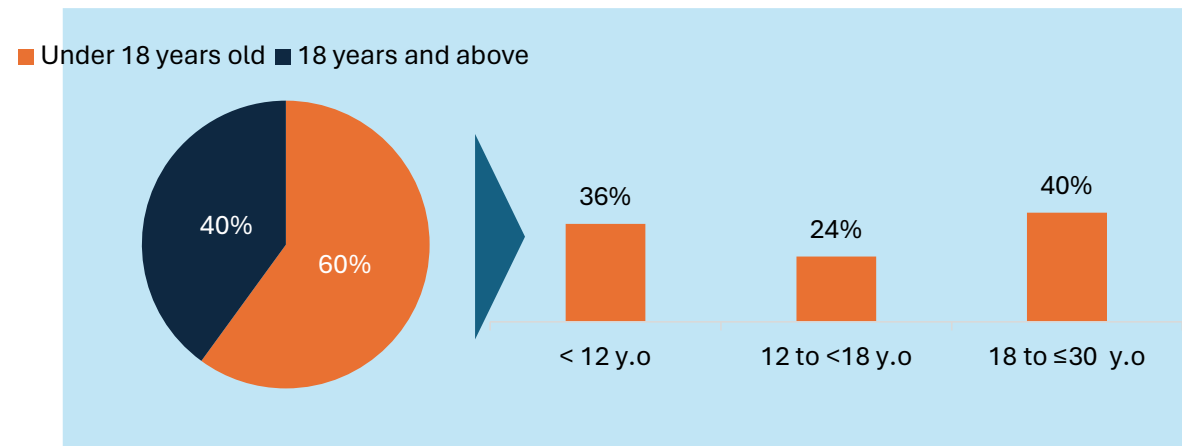
Type of respondent
(n=26)



GD type
(n=26)



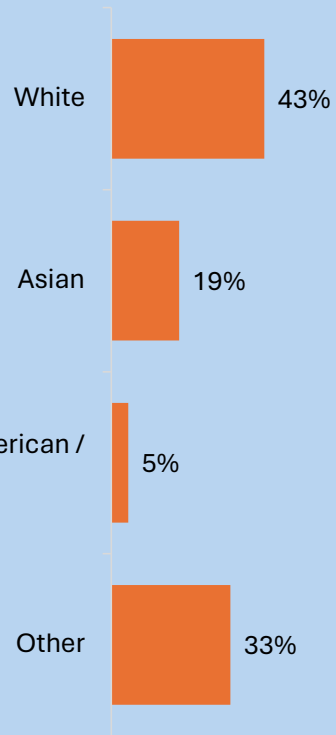
Age of patient at registry entry
(n=25*)



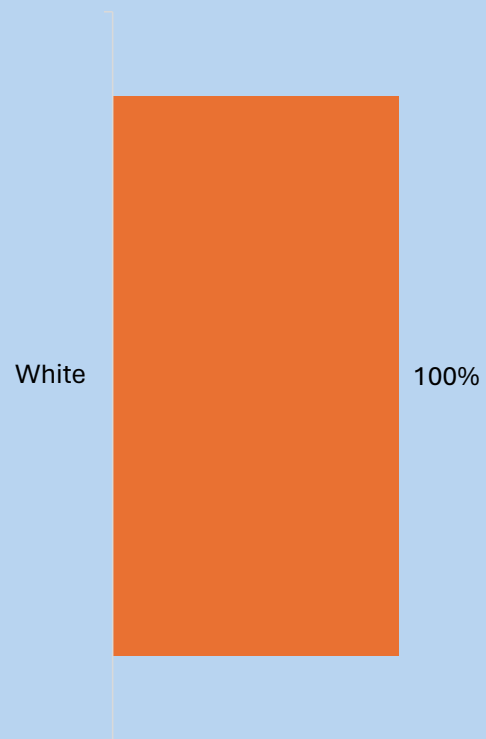
*Results based on 25 patients due to missing data for one patient.

Ethnicity – Overall and by type of Gaucher

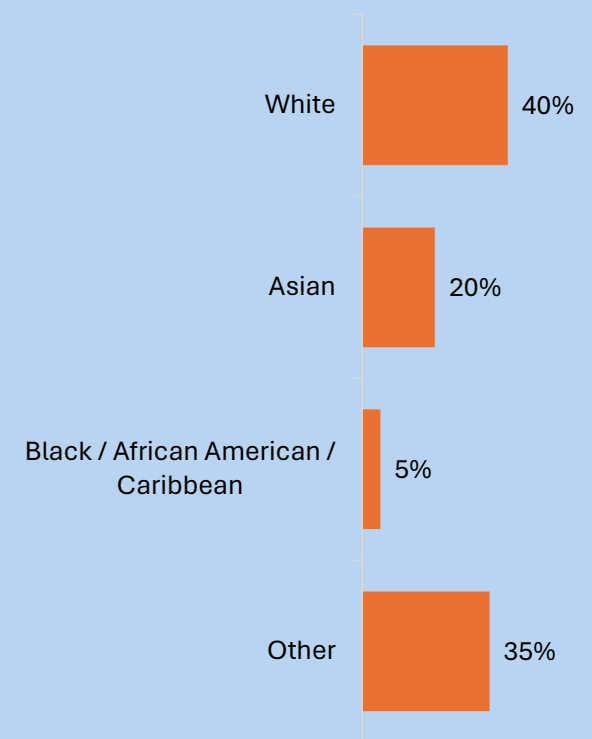
Overall
(n=21*)



GD2
(n=1)

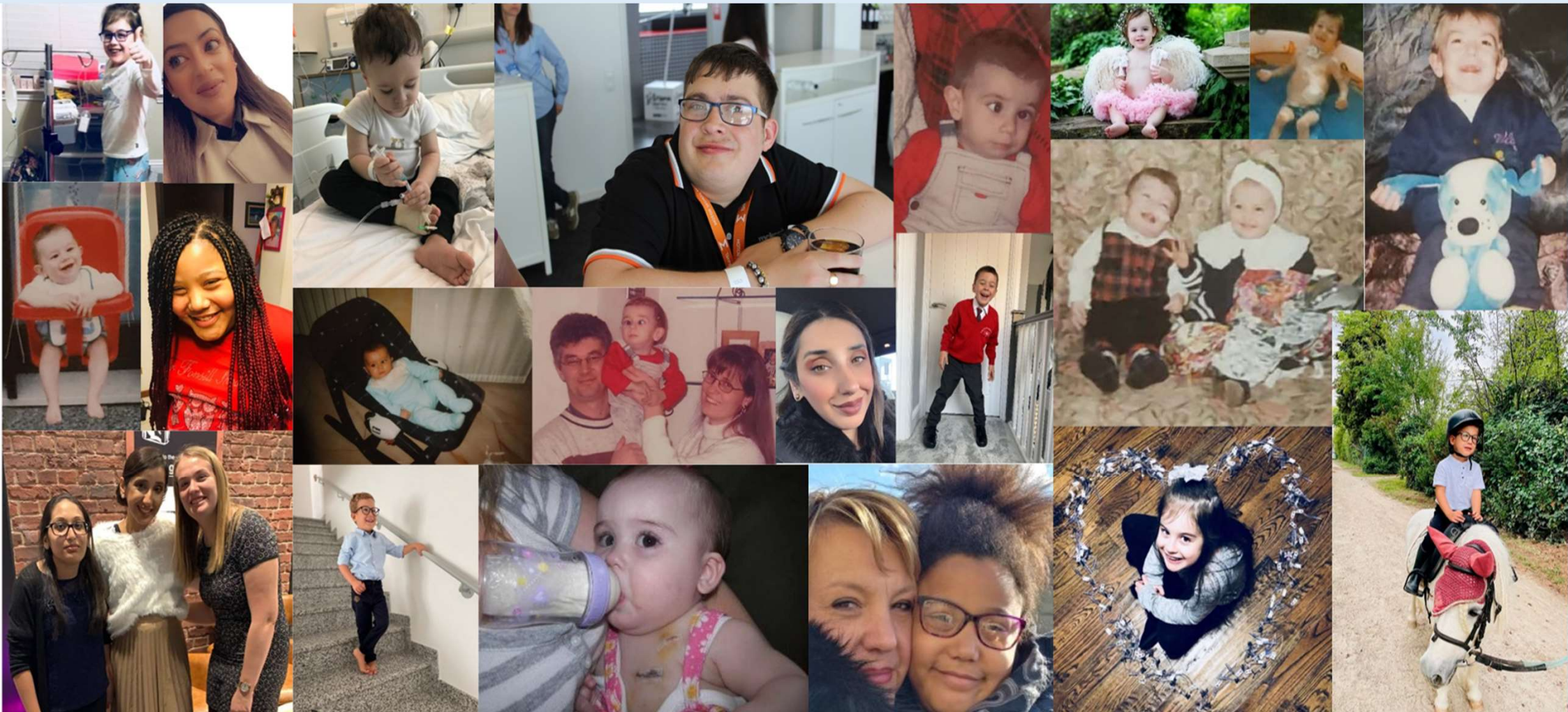


GD3
(n=20)

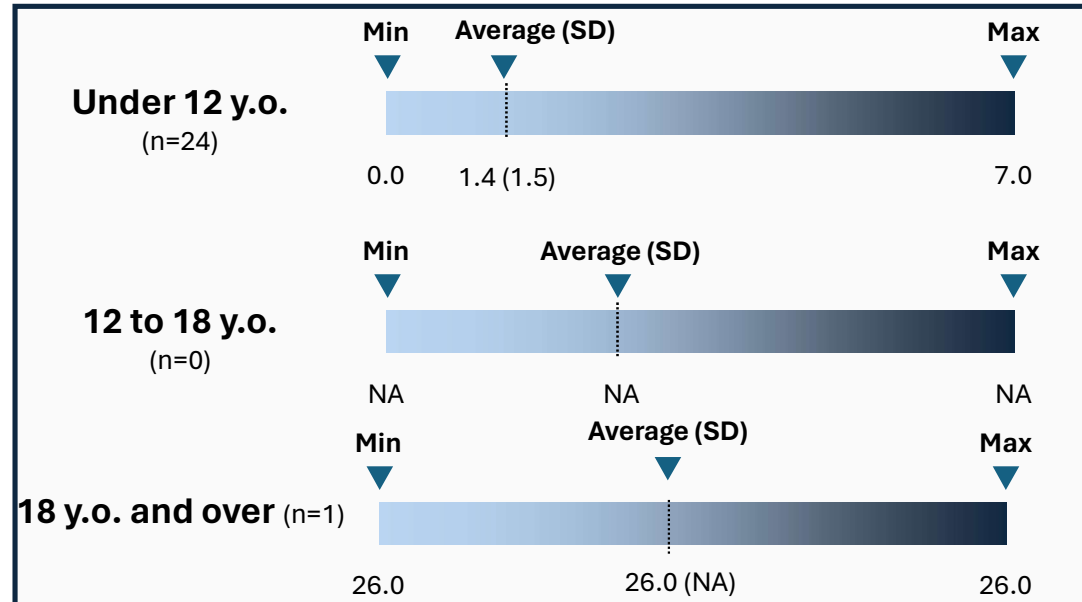
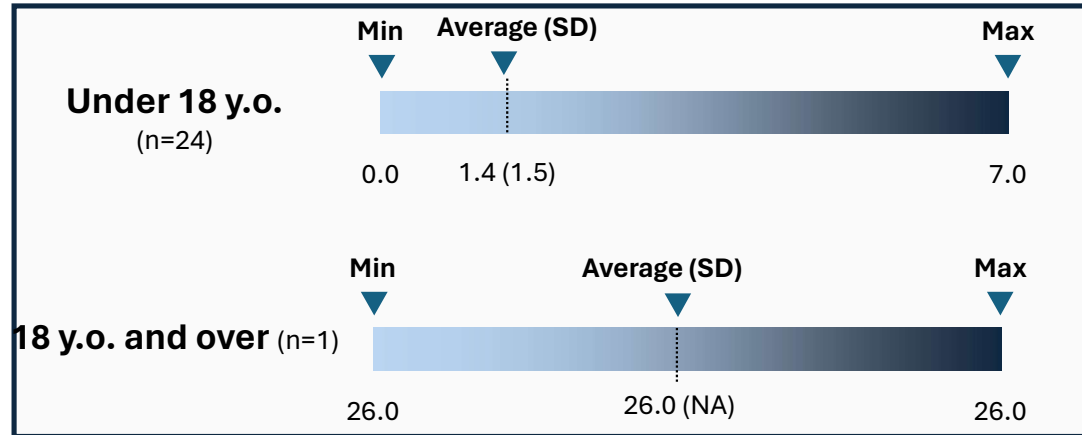
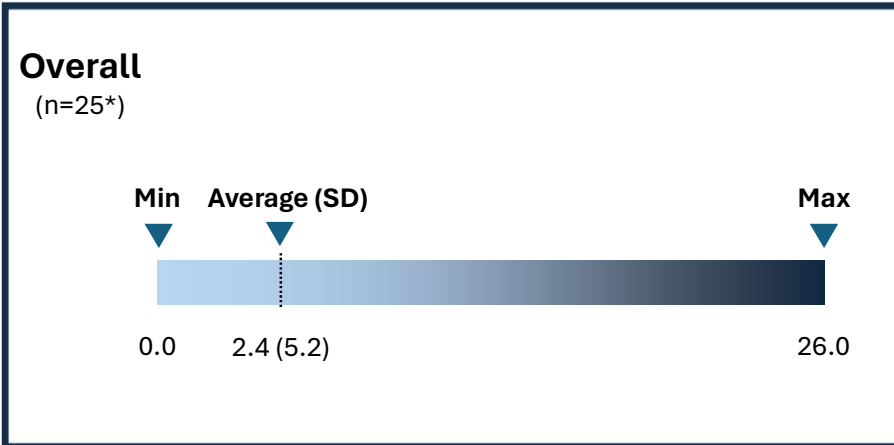


**Results based on 21 patients (due to local regulations, the question on ethnicity was not asked in all countries).

2.1 Age at diagnosis

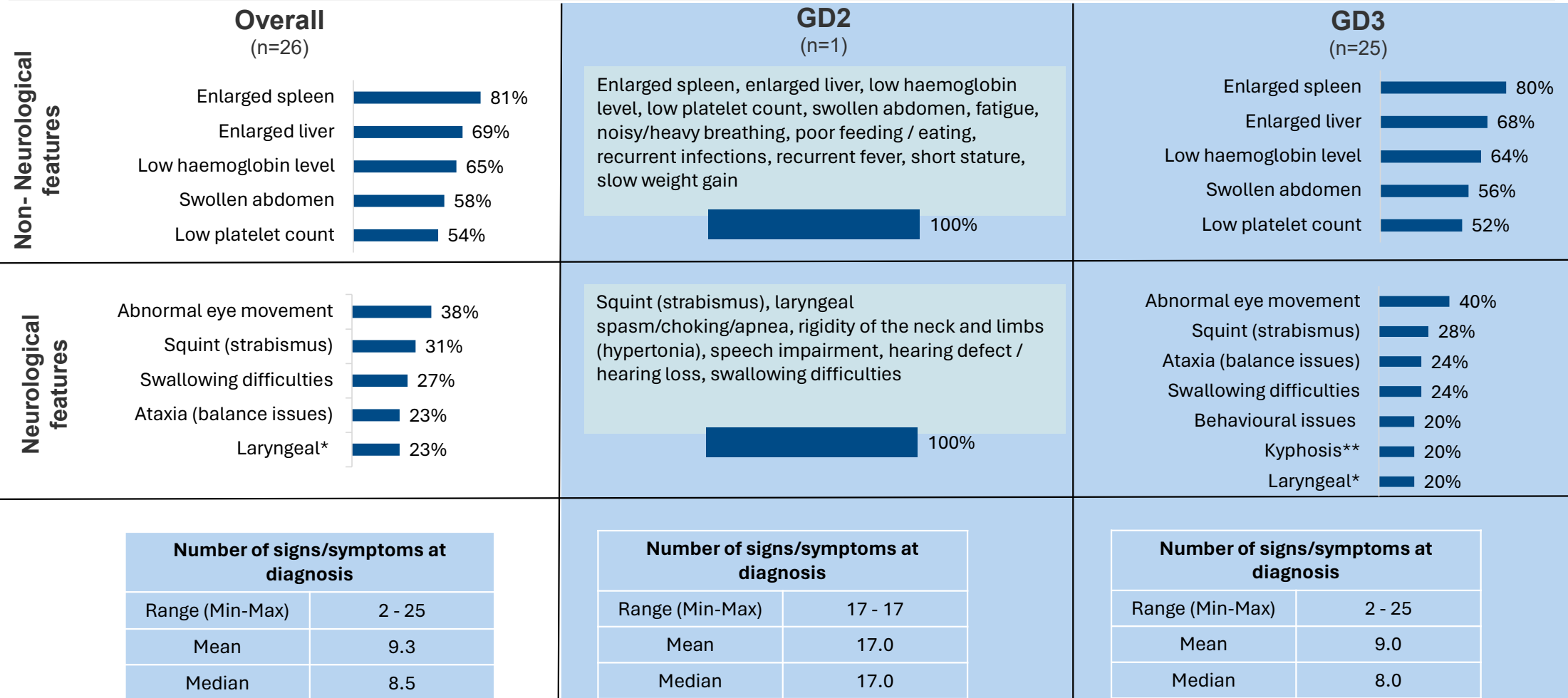


Age at diagnosis – Overall and by age category



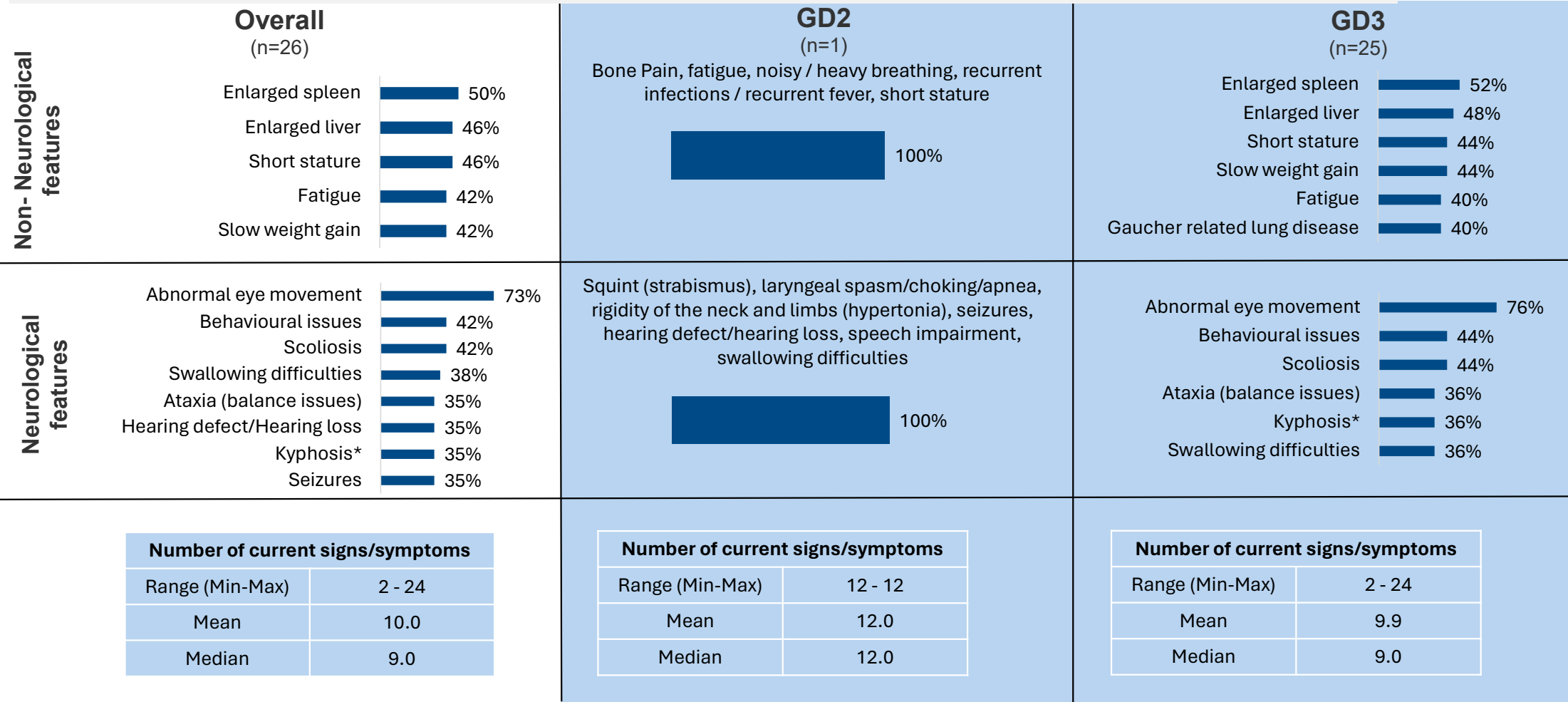
*Results based on 25 patients due to missing data for one patient.

Signs/symptoms at diagnosis (5 most frequently reported) – Overall and by type of Gaucher



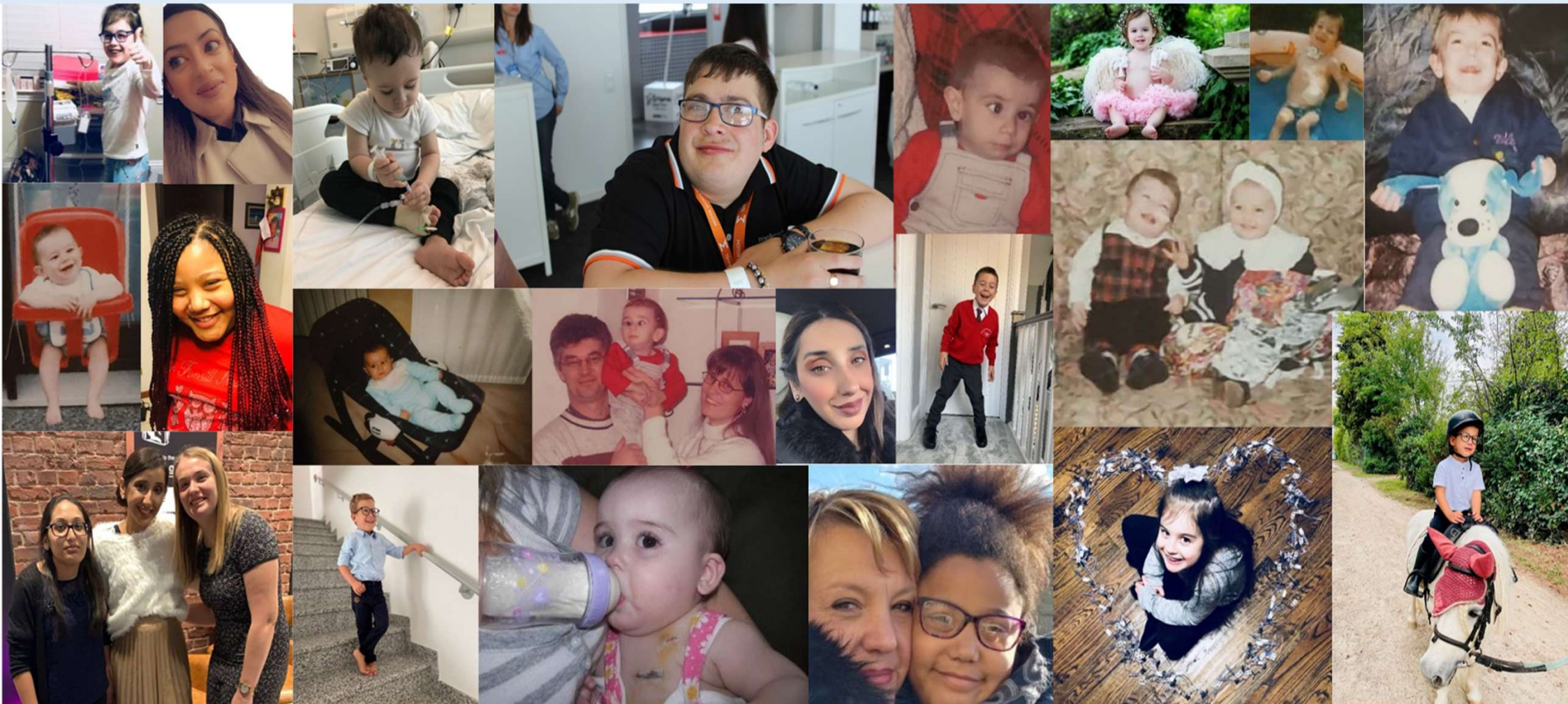
*Laryngeal = Laryngeal spasm/choking/apnea
 **Kyphosis = Kyphosis (abnormal spine alignment)

Current signs/symptoms (5 most frequently reported) – Overall and by type of Gaucher



*Kyphosis = Kyphosis (abnormal spine alignment)

2.4 Treatment and surgeries



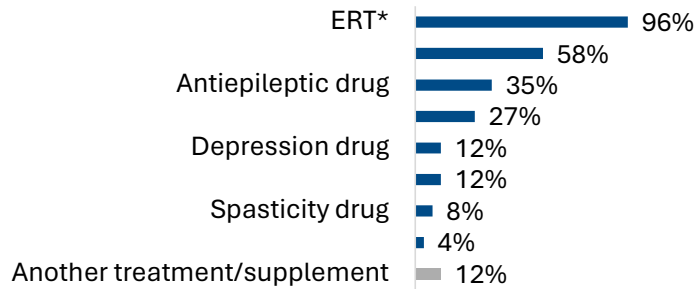
Medical treatment and surgery – Overall and by type of Gaucher

Overall
(n=26)

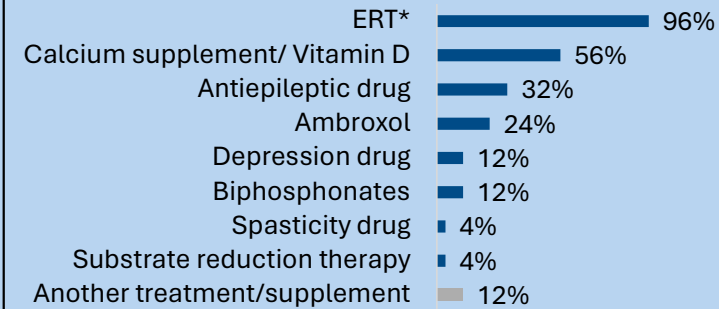
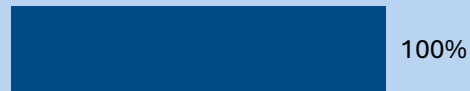
GD2
(n=1)

GD3
(n=25)

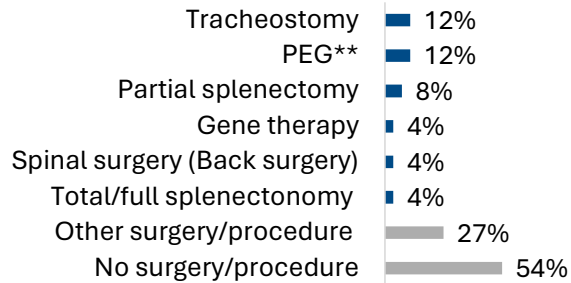
Medical treatment



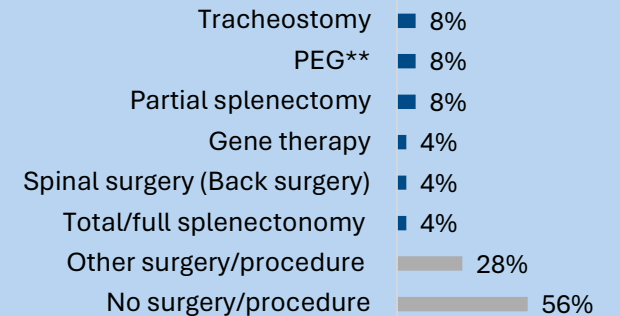
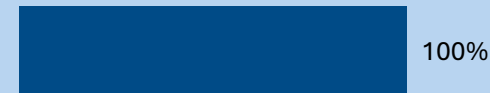
Enzyme Replacement Therapy (ERT), calcium supplement / vitamin D, antiepileptic drug, Ambroxol, spasticity drug



Surgery

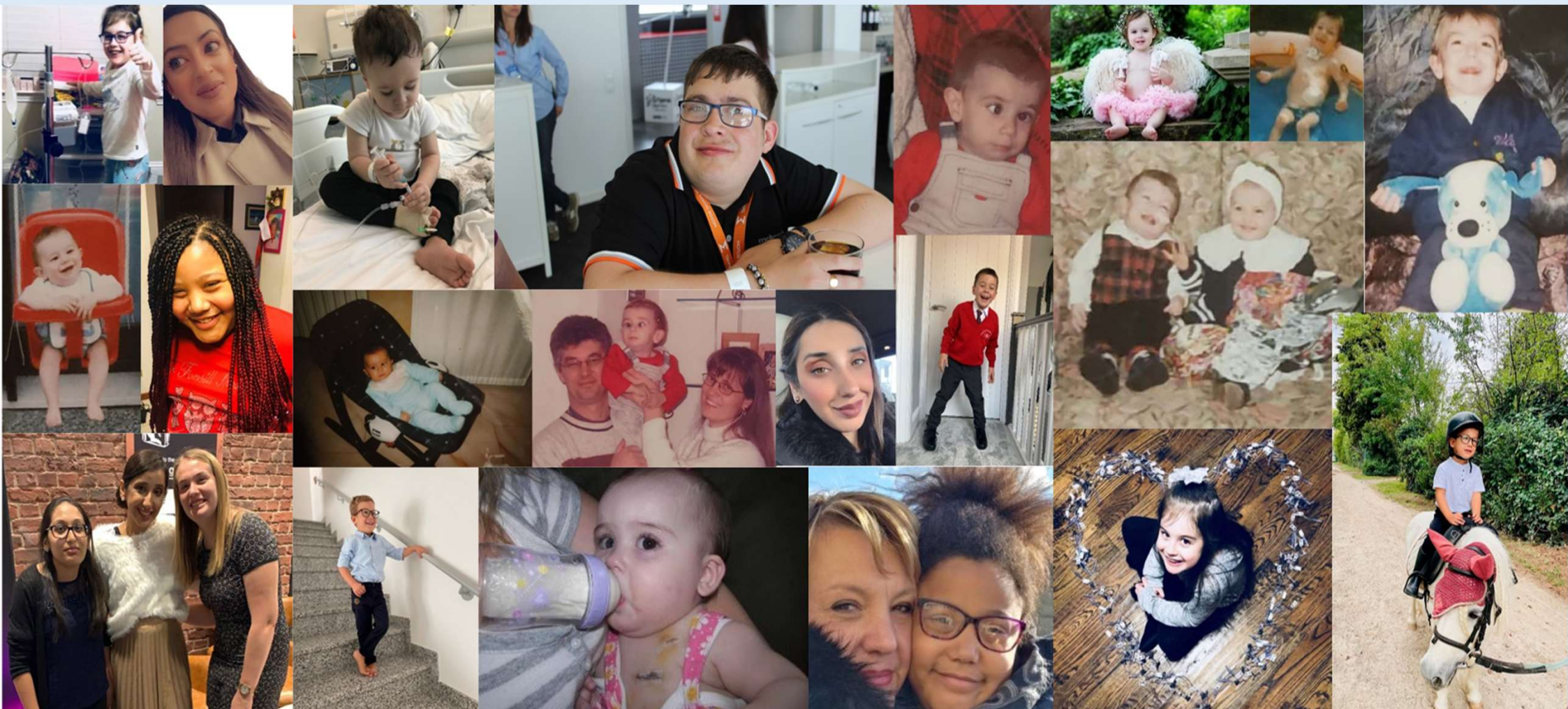


Tracheostomy, feeding tube – Percutaneous Endoscopic Gastrostomy (PEG)



*ERT = Enzyme Replacement Therapy
**PEG = Feeding tube - Percutaneous Endoscopic Gastrostomy

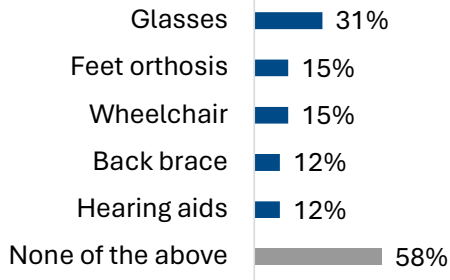
2.5 Medical devices and care services



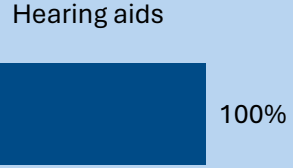
Medical devices and services (5 most reported)– Overall and by type of Gaucher

Overall
(n=26)

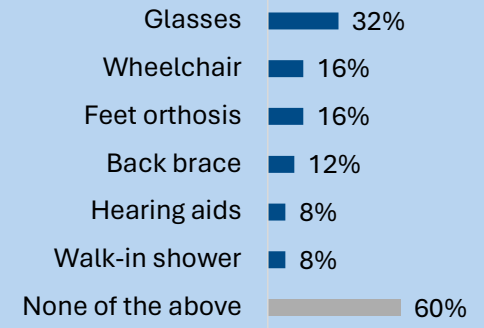
Medical devices currently used



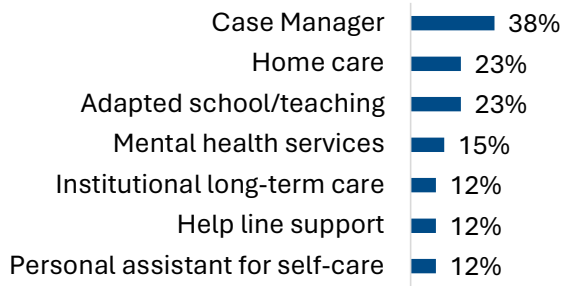
GD2
(n=1)



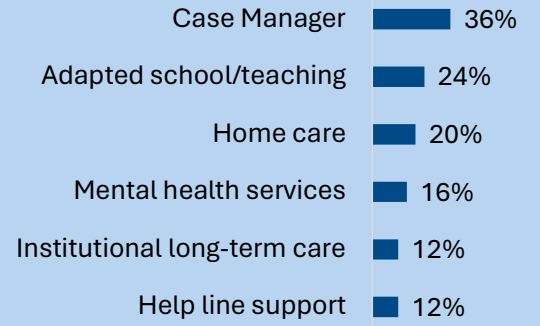
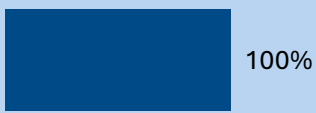
GD3
(n=25)



Services currently used



Home care, case manager, personal assistant for selfcare

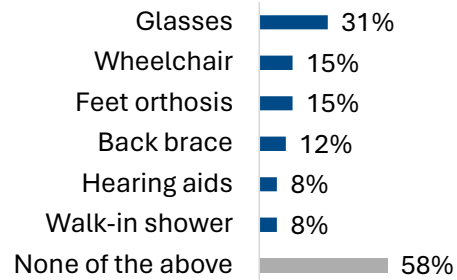


Medical devices and services (5 most reported)– Overall and by type of respondent

Overall

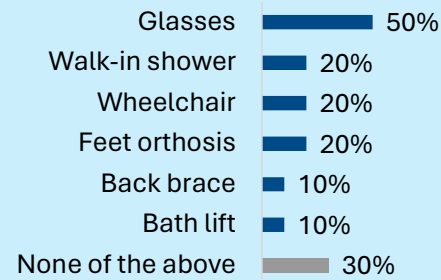
(n=26)

Medical devices currently used



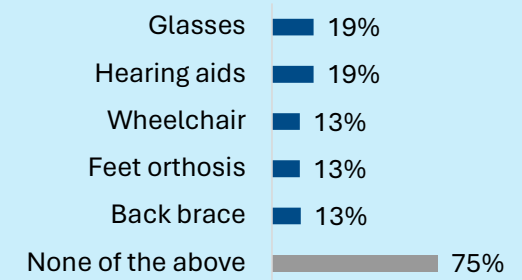
Patients

(n=10)

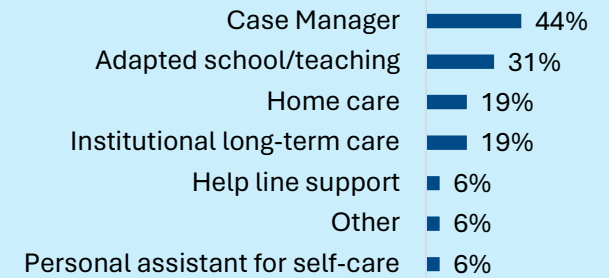
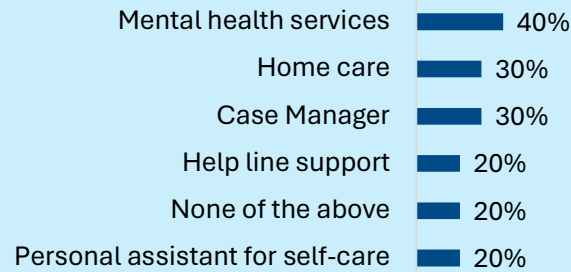
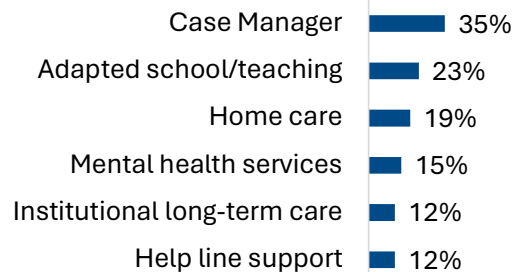


Parents or Caregivers

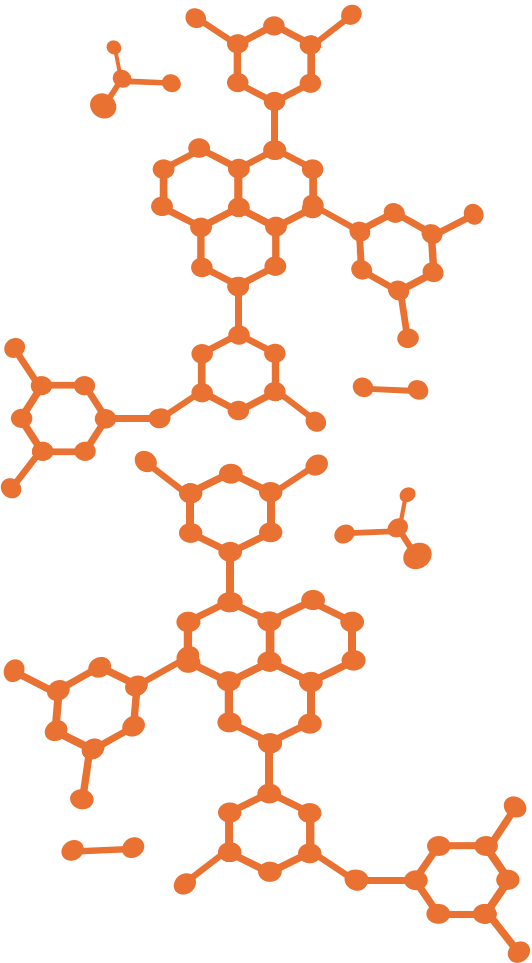
(n=16)



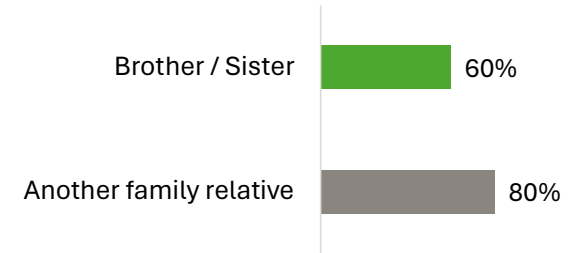
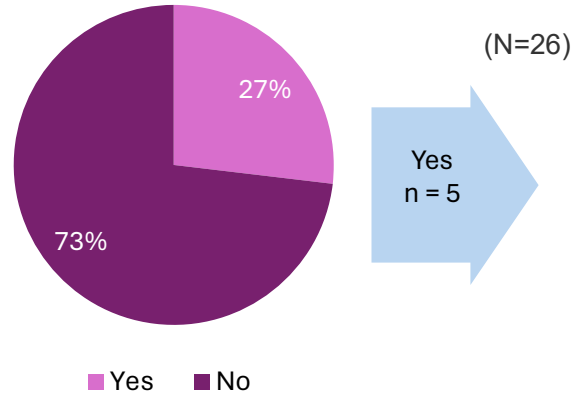
Services currently used



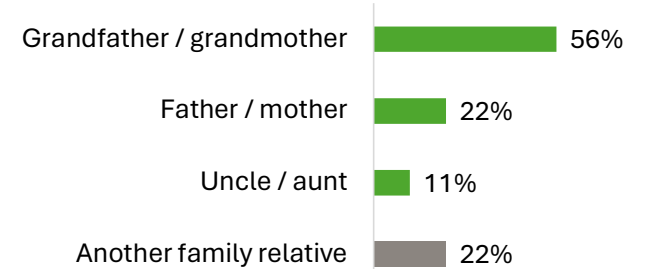
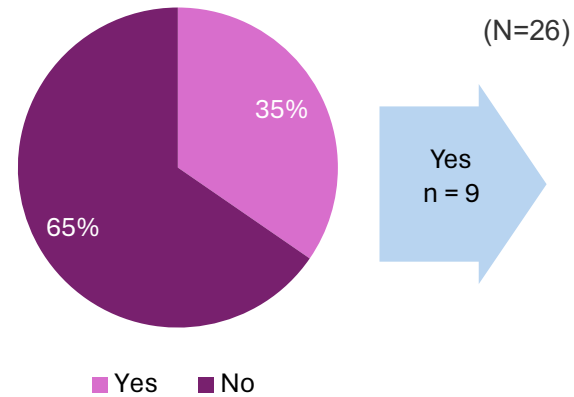
Family history of GD and Parkinson disease - Overall



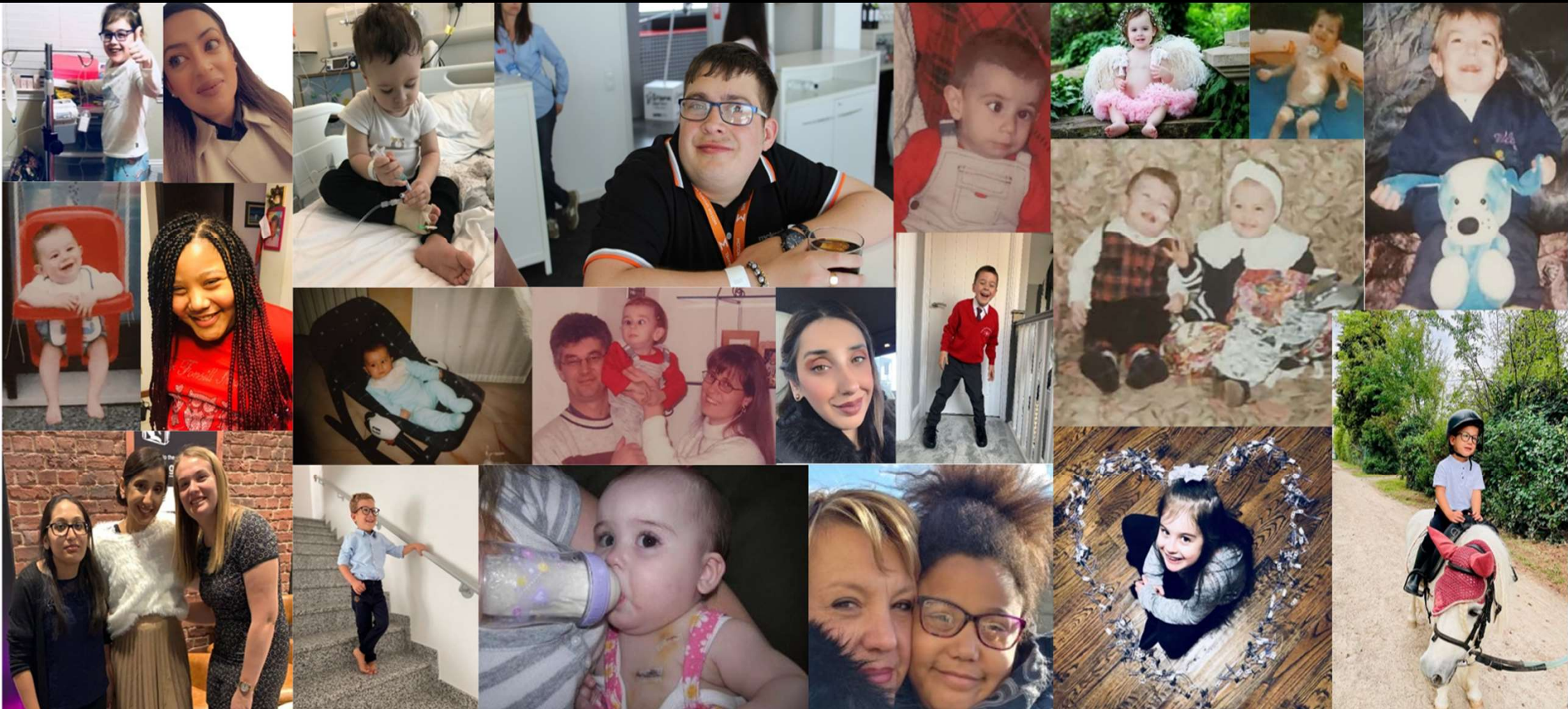
Has anyone else in your family been diagnosed with GD2 or GD3?



Does anyone in your family have Parkinson's disease?

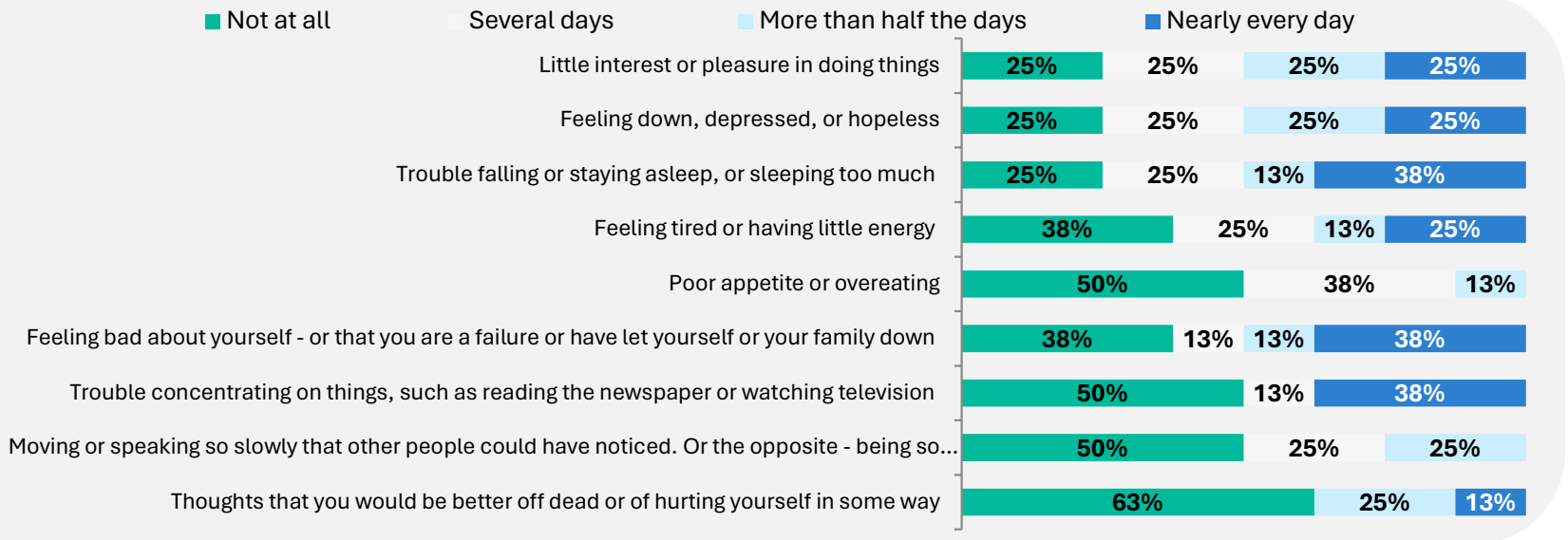


4- Humanistic burden of the disease / Measure of disease severity



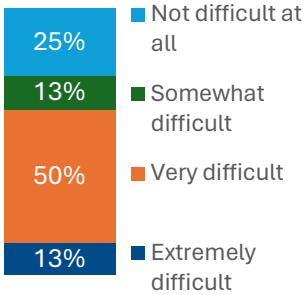
PATIENT HEALTH QUESTIONNAIRE-9 (PHQ-9) – Self-reported by patients

Over the last 2 weeks, how often have you been bothered by any of the following problems? (n=8)

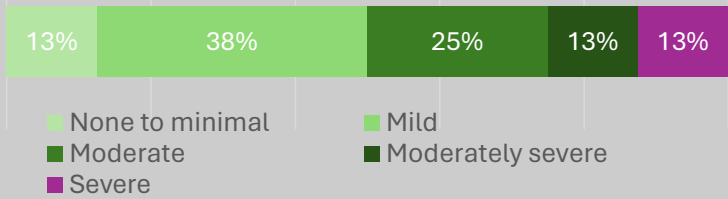


If you ticked any problems, how difficult have these problems made it for you to do your work, take care of things at home, or get along with other people?

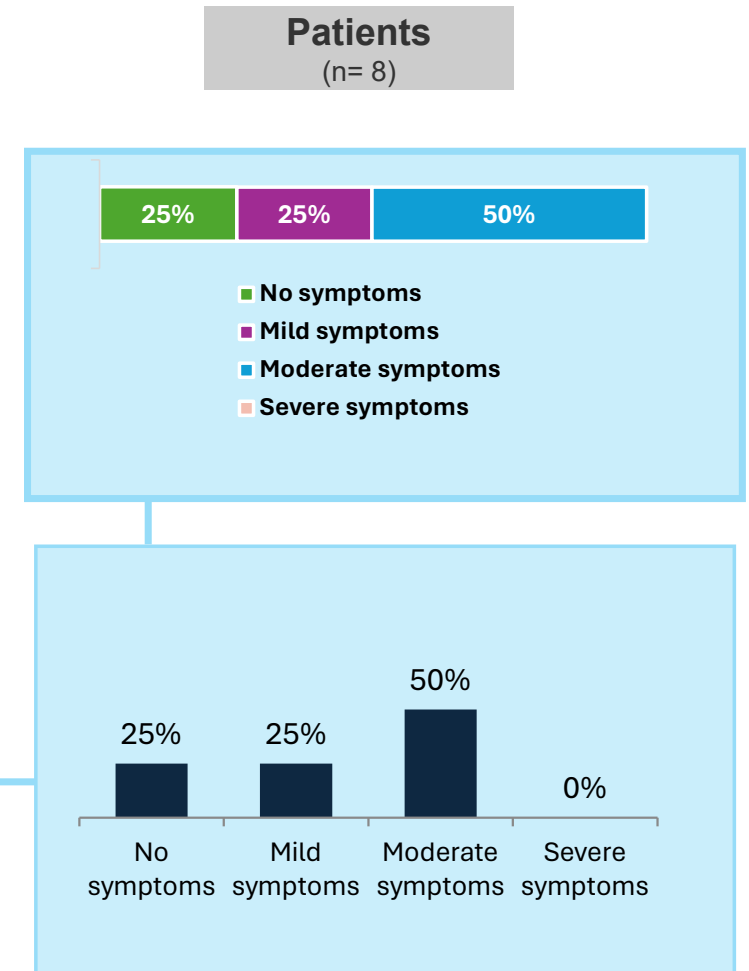
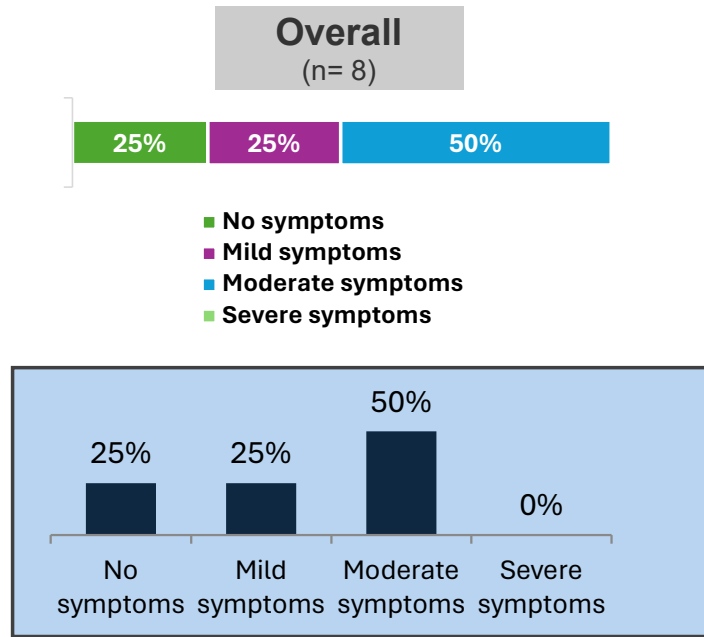
n = 8



Anxiety severity



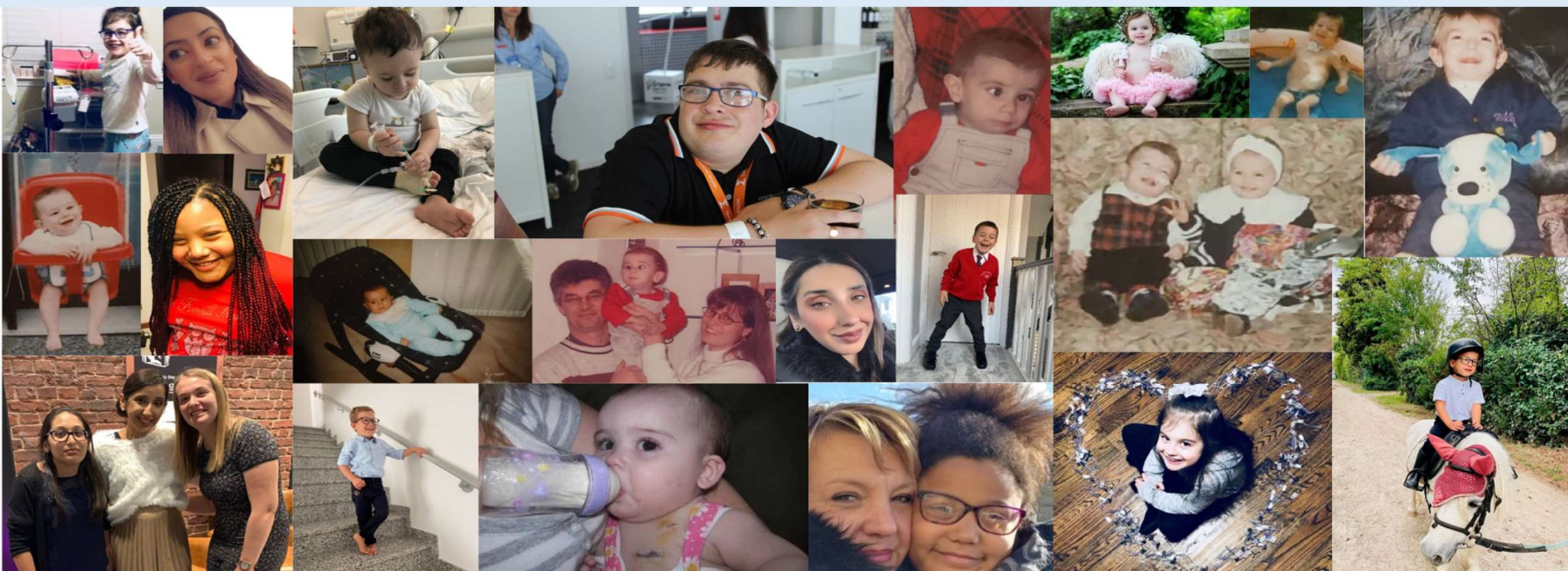
Patient Global Impression of Severity (PGI-S) – Overall and by patients



When thinking about all of the symptoms and physical impacts related to Gaucher Disease that you [your child or the person you care for] may have experienced during the past 14 days, please indicate the one option that best describes how your [your child's/ the person's you care for] symptoms overall have been.

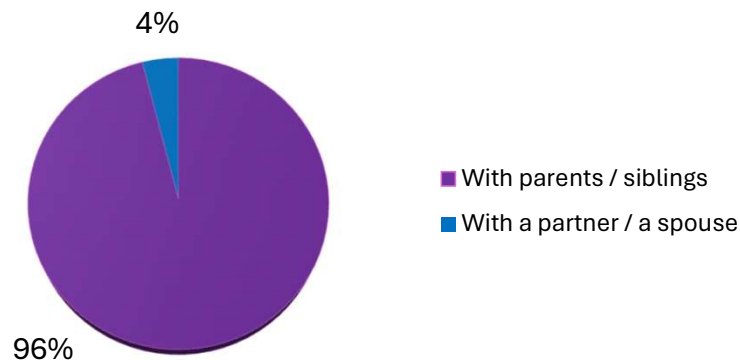
Other patients' characteristics:

- Education
- Employment
- Living arrangement

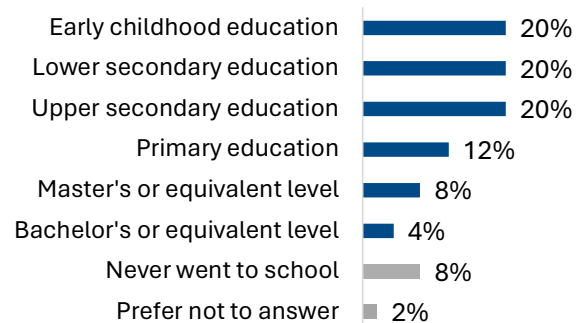


Living situation, education and employment - GD3 overall

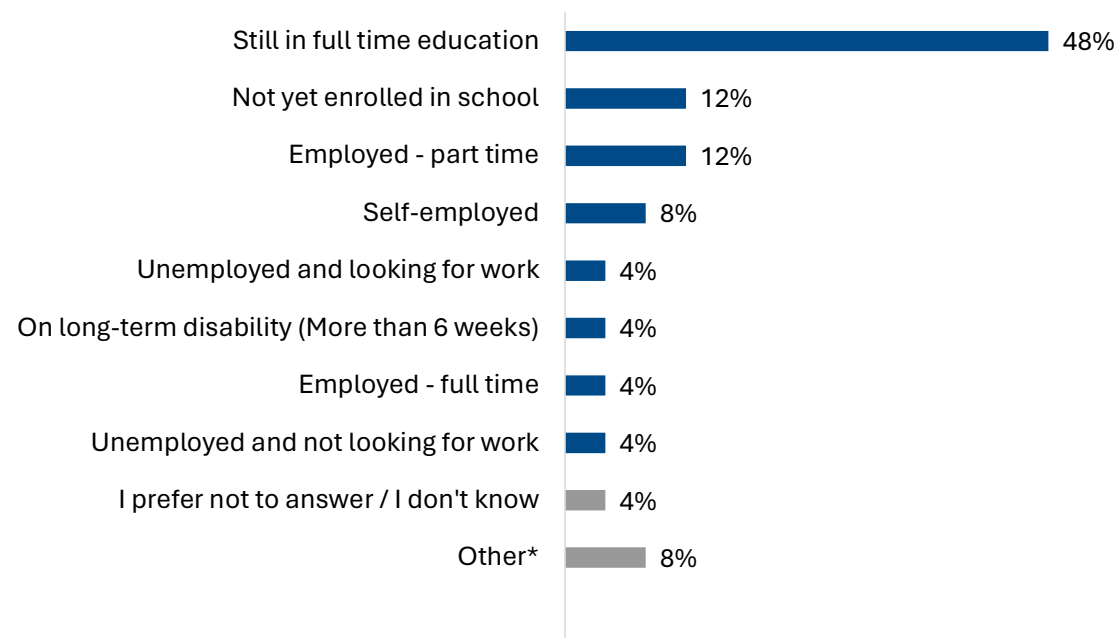
Living arrangement (n= 25)



Education (n= 25)



Employment (n= 25)



*Caregivers specified that the person they cared for was too young to be in employment

Summary of key results

- GARDIAN has data on 26 patients (96% GD3 and 4% GD2).
- Data were provided by patients (38.5%) and parents/caregivers (61.5%).
- There were equal proportions of female (50%) and male (50%) patients.
- Patients had a mean age of 2.4 years at diagnosis and 14.5 years at registry entry.
- The mean number of signs/symptoms at diagnosis was 9.3.
- Neurological symptoms at diagnosis included abnormal eye movements (38%), squint (31%) and swallowing difficulties (27%).
- Non-neurological symptoms most frequently experienced at diagnosis included an enlarged spleen (81%), enlarged liver (69%) and low haemoglobin levels (65%).
- Between diagnosis and registry entry, the prevalence of most non-neurological symptoms and signs decreased.
- Conversely, neurological signs and symptoms increased between diagnosis and registry entry.
- The most common medical treatments involved enzyme replacement therapy (ERT) (96%), calcium/vitamin D supplementation (58%), and antiepileptic drugs (35%).
- Surgeries included tracheostomy (12%), percutaneous endoscopic gastrostomy (feeding tube) (12%), and partial splenectomy (8%).
- Patients frequently used medical devices such as glasses (31%), feet orthoses (15%), or a wheelchair (15%).
- Patients or their caregivers used services such as case managers (38%), home care (23%) or adapted school/teaching (23%).
- Overall, 27% of patients had at least one family member diagnosed with GD2 or GD3, and 35% had a family member diagnosed with Parkinson's Disease.
- According to the Patient Health Questionnaire (PHQ-9), 38% of patients reported that nearly every day over the past 2 weeks, they felt bad about themselves or that they were a failure and had let themselves or their family down.
- A total of 96% of patients lived with their parents/siblings and 4% with a partner/spouse.
- Patients had achieved a secondary (40%) or university level education (Bachelor's or Master's equivalent) (12%).
- Overall, 24% of patients were employed (either full-time, part-time, or as self-employed).

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