

Guideline	Patient-reported outcome measures for patients with Gaucher disease
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Overview	It is now acknowledged that patients' input in health outcome assessment is vital to understanding the impact of diseases and interventions. Though medical technology allows measuring physical, physiological, or biochemical data of the patient, it does not give all the necessary insight and data about the treatment of the disease The panel used the Grading of Recommendations Assessment, Development and Evaluation (GRADE) approach to assess the certainty in the evidence and formulate recommendations (https://www.gradeworkinggroup.org)(1).
Sections	PATIENT-REPORTED OUTCOME MEASURES (PROMS).....2 GAUCHER DISEASE -SPECIFIC PROMS2 GAUCHER SPECIFIC PROMS IN DRUG DEVELOPMENT2 GAUCHER SPECIFIC DIGITAL HEALTH SOLUTIONS3 FUTURE INITIATIVES3 REFERENCES3

Patient-reported outcome measures (PROMs)

Recommendation: The panel suggests integrating patient-reported outcome measures (PROMs) into the routine care of patients with Gaucher disease to improve patient care and the patient-physician relationship.

Remarks:

- Caregiver/observer reported outcomes are also important to detect early signs of neurological pathology, in instances like neuronopathic GD, Parkinson comorbidity, etc. and to monitor the burden of disease on family members.

Details:

Some clinically important data can only be obtained from the patient (or caregivers in some instances). For examples-

1. Physical symptoms not evident to the observer like- bone pain, fatigue and psychological symptoms like- anxiety, depression.
2. The frequency and severity of symptoms using visual analog scale for fatigue, physical weakness, bone pain
3. Nature and severity of disability experienced by the patient
4. The impact of the disease on the daily life of the patient and their family
5. Perception of the patient towards the disease or the treatment given

Patient-reported outcome measures completion prompts patients to reflect on their health and permits them to raise issues with clinicians [1]. The use of PROMs before the clinic visit could support the conversation with the physician. They can also be relevant to self-management.

Gaucher disease -specific PROMS

Recommendation: The panel suggests using Gaucher disease-specific patient-reported outcome measures (PROMs) over the use of generic PROMs.

Details: In the past, PROMs in patients with GD used non-disease-specific tools, including the Short-Form-36, EuroQoL-5 Dimensions, Fatigue Severity Scale, Pediatric Quality of Life Inventory, Symptoms Checklist-90 with limited responsiveness [2]. It is recognized that disease-specific PROMs are better for the aspect of health that is specific to a given disease. Gaucher disease-specific PROMs are currently developed.

A GD1-specific PROM (GD1-PROM) was developed with input from patients, including 15 questions; six Point Verbal Response Scale regarding the last month, and nine Visual Analogue Scales (VAS) from 0-10 regarding the previous week. Although used in one study [3], this PROM is still underdeveloped. An nGD-specific PROM is currently under development, led by the International Gaucher Alliance (IGA).

Gaucher specific PROMS in drug development

Recommendation: The panel suggests the integration of GD-specific PROMs in clinical studies and drug development programs. Preferably globally standardized, to allow synergy between studies, programs, and clinical practice.

Gaucher specific digital health solutions

Recommendation: The panel calls for using and developing GD-specific digital health solutions to improve patient care.

Details: A pilot study of wearable technology in 21 patients (children and adults, type 1 GD and neuronopathic GD) demonstrated the feasibility and patient engagement for such an approach, obtaining a rich dataset useful for proactive clinical care and clinical trial outcome development [4]. Technology has evolved rapidly since then. It now offers remote, passive, easy-to-use solutions for patients to share symptoms, PROMs, standard video-based assessments, and other outcomes with their clinicians as digital biomarkers.

Future Initiatives

- A. The panel acknowledges the need to create an effective Pre- GD Clinic Questionnaire. A short questionnaire can empower the patients and families to make their clinic visits more productive and capture non-GD clinical events that may impact patient care. The stages of development will include literature review and patients and physicians focus groups.
- B. The panel recognizes the increased use of telemedicine in patient care. The integration of telemedicine in patients with GD, its advantages, and barriers, would need to be studied.

References

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