

Additional file 1: Table S1 Topics covered by PROM items in English. Parts 1 and 2 were from the previously published PROM for GD1 (Elstein D, et al. Orphanet J Rare Dis. 2022;17:9), whereas Part 3 was newly developed

Item	Topic
Part 1	Over the past month:
P1-1	restricted education/job
P1-2	restricted activities with friends
P1-3	restricted intimate relationships
P1-4	restricted hobbies/leisure activities
P1-5	concerned about emotional burden to others
P1-6	concerned about risk of bone disease
P1-7	concerned about risk of cancers
P1-8	concerned about risk of Parkinson's disease
P1-9	concerned about financial burden
P1-10	concerned that budget may affect therapy
P1-11	concerned about access to expert physician
P1-12	non-GD problems compared with GD problems
P1-13	general health improvement because of GD medication
P1-14	all medical concerns were GD related
P1-15	current medication treated GD-related concerns
Part 2	Over the past week:
P2-1	GD-related dependence on others
P2-2	GD-related abdomen swelling
P2-3	GD-related fatigue
P2-4	GD-related physical weakness
P2-5	GD-related bone pain
P2-6	GD-related depression

- P2-7 GD-related worries
- P2-8 future with GD
- P2-9 satisfaction with GD medication

Part 3 Over the past week:

- P3-1 difficulty hearing
- P3-2 difficulty seeing
- P3-3 difficulty swallowing food
- P3-4 difficulty speaking
- P3-5 involuntary movement or difficulty moving your arms or legs
- P3-6 epileptic seizures interfering with daily life
- P3-7 pain in the body
- P3-8 feel anxious about above symptoms (No. 1-7)
- P3-9 memory loss
- P3-10 difficulty exercising, studying, or working compared with other persons
- P3-11 feel anxious about continuing GD treatment
- P3-12 feel worried or nervous about going out

Up to this point:

- P3-13 felt physically tired after visiting the hospital or treatment
- P3-14 felt lack of understanding of GD by public service office
- P3-15 lack of social support
- P3-16 participation in GD patient support group

GD: Gaucher disease; GD1: type 1 GD; PROM: patient-reported outcome measure