

Supplementary Table S1. GI-focused medical history to assess the GI health of adult patients with Pompe disease.

- 1) Have you been diagnosed with a gastrointestinal (GI) disease or disorder by a physician? If yes, please list. (Examples include but are not limited to: swallowing dysfunction, esophageal dysmotility, GERD/acid reflux, gastritis, peptic ulcer disease, delayed gastric emptying, gallstones or other gallbladder/biliary disease, irritable bowel syndrome/IBS, celiac disease, Crohn's disease, ulcerative colitis, rectal sphincter dysfunction, fecal incontinence, etc.)
- 2) Have you been evaluated by a gastroenterologist (GI physician)?
- 3) Have you undergone an upper endoscopy? If so, for what reason?
- 4) Have you undergone a colonoscopy? If so, for what reason?
- 5) Have you been diagnosed with TMJ (temporomandibular joint) dysfunction?
- 6) Have you been told that you have tongue weakness by a medical professional (physician, therapist, etc.)? A history of difficulty with chewing, especially meats, any jaw fatigue? Any history of things getting stuck in throat? Difficulty with clearing food with tongue?
- 7) How many stools do you typically have per day?
- 8) If you do not have stools daily, how many stools do you typically have per week?
- 9) Are your GI symptoms worse at any time period of the day? Please state which symptom(s) and its association(s). If the question does not apply to you, please answer N/A.
- 10) Are your GI symptoms temporally associated with meals? Please state which symptoms(s) and its association(s). If the question does not apply to you, please answer N/A.
- 11) Do you take medications to relieve GI symptoms? If so, which medications and how often?
- 12) Do you consider your GI symptoms to be one of your top three symptoms that affects your quality of life?
- 13) From your recollection, did your GI symptoms start bothering you before or after your diagnosis of Pompe disease? Please state how long before/after to the best of your knowledge.
- 14) Has there been a change in your GI symptoms since you started treatment with ERT? Please state what changed and how it changed.
- 15) Is there worsening of GI symptoms after ERT (same day or up to 48 hours after)? Describe.
- 16) Do you have any other comments or concerns regarding GI issues?

Supplementary Table S2. Relationship between raw scores on PROMIS-GI symptom scales and clinical variables in patients with late-onset Pompe disease.

Statistical relationships which were close to being significant or significant		p-value
belly pain (raw score)	patient's age	0.0471*
diarrhea (raw score)	sex (male)	0.0686
swallowing (raw score)	CPK	0.0740
incontinence (raw score)	CPK	0.0619
swallowing (raw score)	FVC % predicted (supine) – inverse relationship	0.0036**
diarrhea (raw score)	FVC % predicted (supine) – inverse relationship	0.0450*
incontinence (raw score)	FVC % predicted (supine) – inverse relationship	0.0264*

*p-value < 0.05, **p-value using Bonferroni correction <0.006.