

Supplementary Table 1. Search Terms for MEDLINE

Search No.	Search Terms
1	Title or abstract: GSDIa OR Glycogen storage disease OR von Gierke's disease OR Pompe's disease OR Cori's disease OR Andersen disease OR McArdle disease OR Hers' disease OR Tarui's disease
2	Title or abstract: Scale* OR Measure* OR Rating* OR Tool* OR Instrument* OR Questionnaire* OR Index* OR Diary* OR Diaries* OR PRO* OR Patient report* OR COA* OR Clinical outcome assessment*
3	Title or abstract: Symptom* OR Quality of life OR QoL OR Health-related quality of life OR HRQoL OR HRQL OR Activities of daily living
4	1 AND 2
5	1 AND 2 AND 3
Filters: Published between 2010 to 2020; English language; Human	

Key: asterisk (*) used as a wildcard or truncation symbol to broaden searches by finding multiple variations of a term or phrase

Supplementary Table 2. Criteria for the Selection of Articles for Full-Text Review

Inclusion Criteria	Exclusion Criteria
<p>Clinician-report or patient-report of a sign, symptom, or impact of GSDIa is included and described in the article; OR</p>	<p>Relevant GSDIa term is included in the title but is not the focus of the article; OR</p>
<p>Article focuses on instruments developed using clinician or patient input to measure signs, symptoms, or impacts of GSDIa.</p>	<p>Article focuses on unusual presentations of GSDIA (i.e., case studies) or specific populations (e.g., GSDIa in pregnant women); OR</p>
	<p>Article is a systematic review containing and citing information included in other articles (i.e., repetitive information); OR</p>
	<p>Article reports on purely objective, physiological measures of GSDIa (e.g., medical imaging).</p>

Supplementary Table 3. Stability of Daily Total Scores Across 7-Day Periods During Screening

Daily score	Visit	N	Time points	Estimate	L95	U95
Symptoms Total	Baseline - 14 Days to - 8 Days	15	7	0.95	0.9	0.98
Daily Impacts Total		5		0.99	0.97	1
Sleep Impacts Total		16		0.93	0.86	0.97
Symptoms Total	Baseline - 7 Days to - 1 Day	26	7	0.98	0.97	0.99
Daily Impacts Total		14		0.99	0.97	1
Sleep Impacts Total		26		0.95	0.92	0.97

Key: N, number of participants with complete data across time points; ICC, intra-class correlation coefficient; L95, lower bound of 95% interval for ICC (A,K); U95, upper bound of 95% interval for ICC (A,K).

Supplementary Table 4. Changes in Score Reliability Due to Missing Daily Scores Using ICC (A, K) Estimates from the Last 7-Day Period of Screening

7-Day Average Score	Timepoint	7 days	6 days	5 days	4 days
Symptoms Total	Baseline -7 Days to -1 Day	0.98	0.98	0.97	0.97
Daily Impacts Total		0.99	0.99	0.99	0.98
Sleep Impacts Total		0.95	0.94	0.93	0.92

Key. ICC, intra-class correlation coefficient

Supplementary Table 5. Minimum Detectable Change and Significant Within-Patient Change Thresholds for the GSD Domain Scores from the Last Seven Days of Screening

MDC index	Estimate	Significant within-patient change threshold
Symptom Total Score		
SEM	0.84	1.65
SEE	0.83	1.63
SEP	1.19	2.33
RCI	1.19	2.33
Sleep Impacts Total Score		
SEM	0.58	1.14
SEE	0.56	1.10
SEP	0.80	1.57
RCI	0.81	1.59
Daily Impacts Total Score		
SEM	1.06	2.08
SEE	1.05	2.06
SEP	1.49	2.92
RCI	1.50	2.94

Key: SEM, standard error of measurement; SEE, standard error of estimation; SEP, standard error of prediction; RCI, reliable change index

Note: The reliability assumption used to estimate these indices was the ICC (A, K) estimated using the daily scores over the last seven days of screening. Bolded values represent the upper and lower bound of the MDC range.

Symptom Total Score ICC (A, K) = 0.98

Sleep Impacts Total Score ICC (A, K) = 0.95

Daily Impacts Total Score ICC (A, K) = 0.99