**Supplementary Material 4**

**Full-text Screening Instrument**

**Last name of first author:**

**Year of publication:**

**1. Is the study published in English?**

YES NO

**2. Does the study include patients who have at least one disease of interest?\***

YES NO

Diseases of interest include the following lysosomal storage disorders:

* Gaucher, Type I (non-neuropathic)
* Fabry disease or Anderson-Fabry Disease, Hereditary Dystopic Lipidosis, Angiokeratoma Corporis Diffusum or Alpha-Galactosidase A Deficiency
* Niemann-Pick (Type B), or non-neuronopathic Niemann-Pick
* Pompe disease, or Glycogen Storage Disease Type II, or Acid Maltase Deficiency, Generalized Glycogenosis, Type 2 Glycogenosis
* Mucopolysaccharidoses Type I (MPS I):
  + MPS I H, or Hurler syndrome, or α-L-iduronidase deficiency, or gargoylism
  + MPS I S, or Scheie syndrome
  + MPS I H-S, or Hurler- Scheie syndrome
* Mucopolysaccharidoses Type II (MPS II), or Hunter Syndrome

\*Patients with other diseases may be included in the sample, but as long as the study includes patients with one of the above disease, the study should be included.

**3. Is the study design eligible?**

YES NO

We are EXCLUDING the following types of articles: study protocols, letters, editorials, literature reviews, systematic reviews, and qualitative research

We are interested in PRIMARY and QUANTITATIVE studies. Eligible study designs include, but are not limited to:

* Non-experimental (cross-sectional study, case study, case series, case-control, cohort)
* Experimental (uncontrolled, controlled or randomized control trial)
* Patient-reported outcome instrument development and/or testing (in context of our rare diseases of interest)

**4. Does the study use at least one eligible outcome measure?**

YES NO

An eligible outcome measure would comprise information reported by a patient or a caregiver (parent or guardian). It would take the form of an instrument that is either administered as part of an interview or as a stand-alone measure. Eligible instruments will contain items (or questions) that “tap into the patient experience,” by capturing (either through all or some items) the following information:

* Symptoms (pain, headaches, sleeplessness, etc.)
* Physical, mental/emotional, or social functioning
* Disease-specific conditions
* Satisfaction with treatment
* Overall sense of well-being (global impression score)
* Utility

If the instrument consists (or seems to consist) of patient-reported or caregiver-reported AND clinician-reported items, please include the study and make a note in the comments field.

We are interested in instruments that capture the above information, irrespective of:

* Whether it has been previously validated or it was an ad hoc instrument developed by the study investigators
* Whether it produces a quantitative score (overall or by domains) or not.

We are limiting our inclusion of the untested PRO instruments to the following:

1. instruments that include 3 or more response options (i.e., they are conceivably responsive to change),
2. instruments that dichotomize responses (e.g., Do you have pain? yes or no) will only be included if they have at least 20 items, and
3. instruments that combine dichotomous response options (e.g. yes or no) and 3-response options (e.g. mild, moderate, severe).

With respect to case reports, authors often describe that “Patient X reported pain….” Unless the authors provide information on how they measured pain, i.e. details of the instrument, please do NOT include the study.

Below are examples of eligible and ineligible measures:

Eligible measures

* Brief Pain Inventory
* Children’s Depression Inventory
* Chronic Respiratory Questionnaire
* Dizziness Handicap Scale
* EuroQol (EQ-5D)
* Fabry-specific Pediatric Health and Pain Questionnaire
* Fabry Disease Severity Scoring System (DS3)
* Fatigue Severity Scale
* Fibromyalgia Impact Questionnaire
* Hopkins Symptom Checklist 90 for Anxiety
* KINDL
* Medical Outcomes Study health distress scale (modified)
* Rotterdam Handicap Scale
* Short Form McGill Pain Questionnaire
* Short Form-36
* VAS or numeric rating scale for pain
* World Health Organization Quality of Life questionnaire

Ineligible measures

Measures such as the Brooke score, the Vignos score, the Abnormal Involuntary Movement Scale (AIMS), the Mullen Scales of Early Learning, the Modified Rankin Scale, the FLACC, and the Modified Medical Research Council are ineligible because they are SOLELY completed by clinicians.

Measures such as the Zimran’s severity score index, the Severity Score Index for Gaucher, and the Hermann Score are ineligible because they are SOLELY made up of biological or physiological outcomes.

Cognitive, intelligence, and personality measures are ineligible because they are used to diagnose, and they do not “tap into the patient experience.” These include:

* Minnesota Multiphasic Personality Inventory
* Vineland Adaptive Behavior Scales
* Wechsler Adult Intelligence Scale-III, Woodcock Johnson Tests of Cognitive Ability - Third Edition
* Wechsler Intelligence Scale for Children-Third Generation
* Wechsler Preschool and Primary Scale of Intelligence

**5. Is the paper eligible for data extraction? (Answer must be YES for questions #1-4)**

YES NO