Capturing symptoms and their impact in Fabry Disease



ABOUT THIS RESEARCH

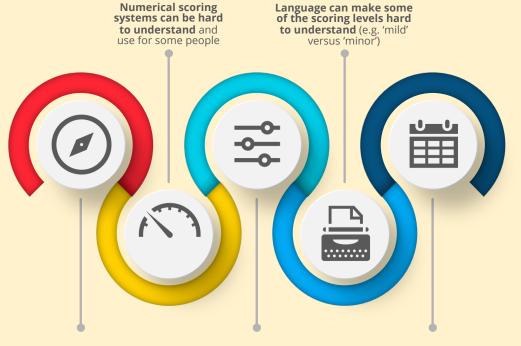
In December 2020 and March 2021 people with Fabry Disease, representing five European countries shared their perspectives on standard questionnaires often used to capture the experiences of people with Fabry Disease and the ideal profile of new tools, designed to more Fabry Disease symptoms and impacts.

hosted by the Fabry International Network

Challenges with current questionnaires

Participants reviewed three standard questionnaires: The Brief Pain Inventory (short form); the Gastrointestinal Symptom Rating Scale; and the EQ-5D-5L

Below are the common themes that emerged:



Ouestionnaires lack guidance and examples to help people understand how to fill them in

Rating scales with five **levels were preferred**. Seven- and ten-level scales can lead to confusion

Inappropriate time period **covered in questionnaires.** Many only cover one day or one week but are filled in annually

Review of specific tools

Brief Pain Inventory (short form)

- Inappropriately covers only one day
- 10-point numerical scale is confusing
- Missing details on the type, duration and causes of pain



Gastrointestinal Symptom Rating Scale

- Covers all the main symptoms experienced by people with FD
- The descriptive rating scale is easy to understand
- The seven-level rating scale could be reduced to five-levels



What could a new tool for Fabry Disease look like?

Members of the Patient Forum recommended that new tools be created that capture the experiences of Fabry Disease in a way that respects the nature, frequency and duration of the symptoms experienced:

EQ-5D-5L

- Too generic to capture the experiences of FD
- Missing important symptoms such as Fabry Fog and fatigue
- Some domains should be split (anxiety and depression are not the same)
- The question on 'your health today' seen as inappropriate and difficult to answer





in real time where possible using diaries, smartphone apps and similar techniques

RECALL OF SYMPTOMS

LANGUAGE TO DESCRIBE **SYMPTOMS**







Consider diagram representation of all symptoms in a tool for people to combine the range of symptoms they are experiencing

SYMPTOMS ARE LINKED TOGETHER



CONNECT **EMOTIONS TO SYMPTOMS**

Relate the emotional impact to the symptoms that people experience. These symptoms directly impact mood



Important aspects of symptoms to consider

Hard to accurately describe Highly impacts daily activities Diverse locations of pain Pain is linked to other FD

GI SYMPTOMS

Defined in part by foods avoided Wide range of symptoms Impacted by stress Hard to discuss with others Impacts many daily activities

FATIGUE

EQ5D

"Like being an empty battery" Has a high impact on daily life Causes frustration and anger Hard to separate from other symptoms

FABRY FOG

Temporary loss of memory and concentration lapses Linked to fatigue and hard to define Moments of terror and isolation

EMOTIONS

Negative emotions caused by other Fabry Disease symptoms Feel isolated due to the hidden nature of the disease





