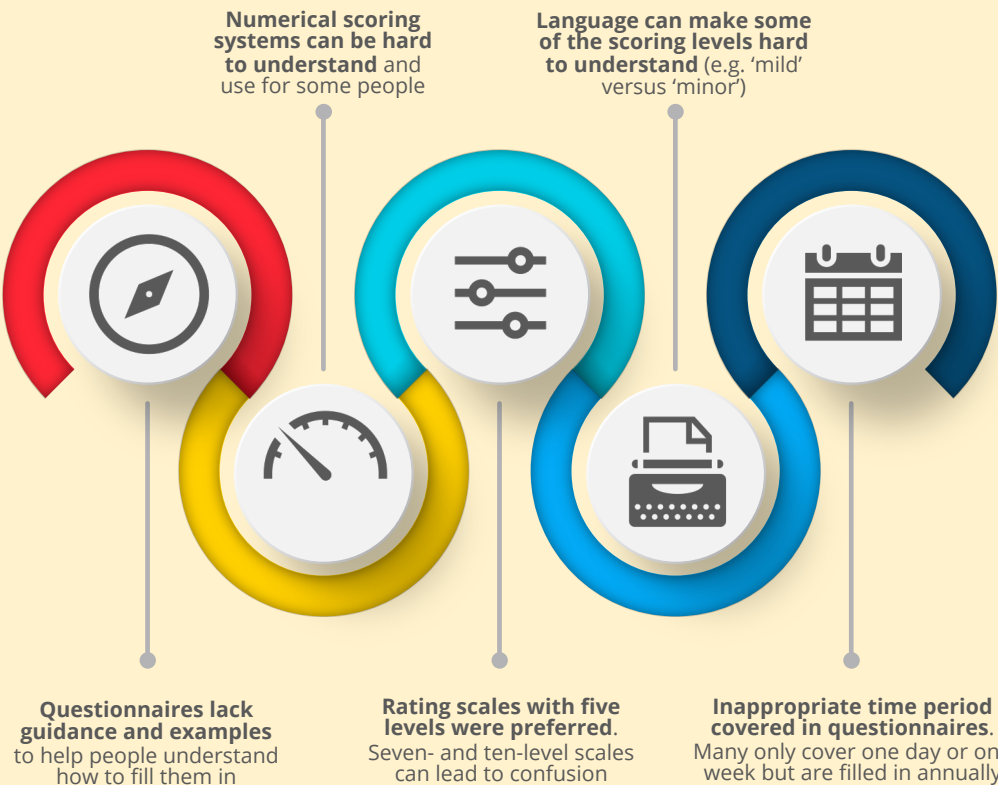


Capturing symptoms and their impact in Fabry Disease

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:



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:



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 accurately record the Fabry Disease symptoms and impacts.

The two Patient Forum meetings were convened by Chiesi Rare Diseases and hosted by the Fabry International Network

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

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



Important aspects of symptoms to consider

PAIN

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

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

"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