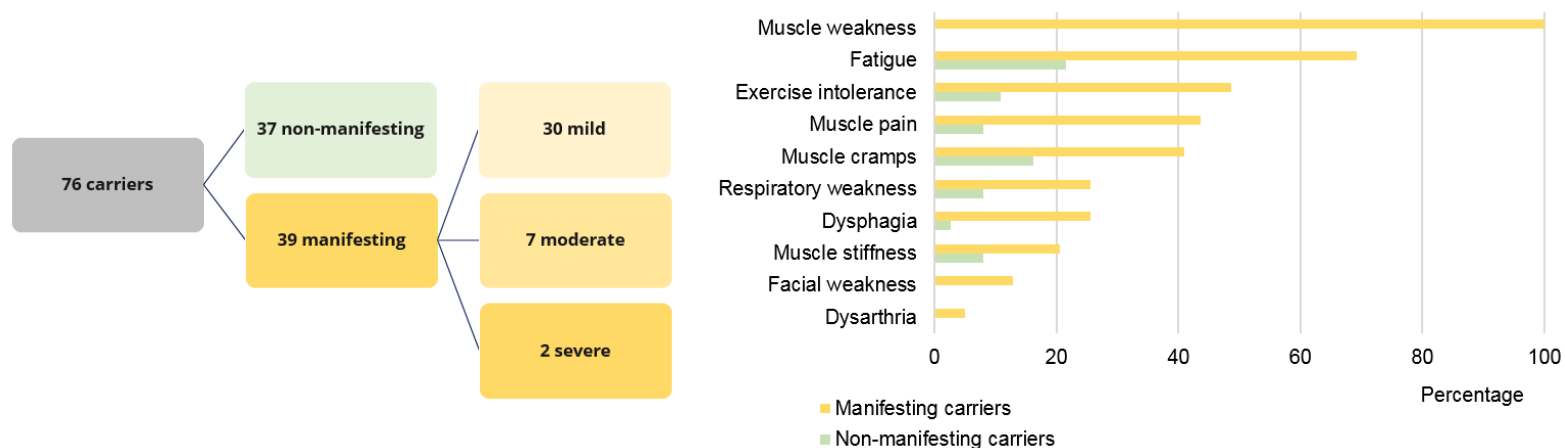


Outcomes of questionnaire study on symptoms in female MTM carriers.

In 2020, a questionnaire study was conducted into myotubular myopathy (MTM) in female carriers. The aim of this study was to find out how many female carriers have muscle complaints, how severe these complaints are and what the other most common complaints are. Also, we wanted to know what the influence of the complaints is on daily life.

For this research, the Netherlands and Germany collaborated with the United Kingdom, and female MTM carriers from different countries were invited to participate. Ultimately, 76 women participated in the study and completed the questionnaires.

About half of these carriers had muscle weakness (also called 'manifesting' carriers), the other half had no muscle weakness (also called 'non-manifesting' carriers). The majority of the manifesting carriers could walk independently (mild), but 7 of these carriers required a walking aid (moderate), and 2 carriers required a wheelchair (severe). In addition to muscle weakness, there was a wide variety of other muscle complaints that occurred, including: fatigue, exercise intolerance, muscle pain, and muscle cramps. These complaints occurred in both manifest and non-manifest carriers, but more often in manifest carriers. In addition, manifest carriers generally suffered more from fatigue, were less able to perform daily activities, and had a reduced physical quality of life. Pain was more frequent in the manifesting carriers, but the severity of the pain was no worse than in the non-manifest carriers.



Based on the results of this study, we can conclude that muscle complaints in female MTM carriers are more common than previously assumed. Mapping out the variation of complaints is an important step towards greater recognition and acknowledgement of these complaints in carriers. This research was performed to better understand this, and further research could provide us with even more information. This is important to be able to provide better care, but also to allow female carriers with complaints to participate in studies into a drug for MTM.

The outcomes of this research have been published in the scientific journal *Neurology*.

Drs. Stacha Reumers

Dr. Frederik Braun