

Which in-clinic assessments are helpful to measure function in individuals with LGMD2A/R1 (Calpainopathy)

Meredith K. James PT PhD^{1*}; Lindsay Alfano PT^{2,3*}; Megan Iammarino PT⁶; Natalie Reash PT²; Chris Steiner PT²; Audrey Beale²; Melissa Smith PT²; Stephanie Hunn PT⁷; Shelley Mockler PT⁸; Heather Hillsden¹; Dionne Moat PT¹; Jassi Sodhi PT¹; Karen Wong PT¹; Emma Grover PT¹; Emma Robinson PT¹; Anna G. Mayhew PT PhD¹; Michelle Eagle PT PhD⁴; Volker Straub MD PhD¹; Michela Guglieri MD¹; Chiara Marini Bettolo MD PhD¹; Robert Muni Lofra PT PhD¹; Jordi Diaz-Manera MD PhD¹; Linda Lowes PT PhD⁵ *Joint first author

Affiliations : 1 John Walton Muscular Dystrophy Research Centre, The Newcastle upon Tyne Hospitals NHS Trust and Newcastle University, UK 2 Abigail Wexner Research Institute at Nationwide Children's Hospital, USA 3 The Ohio State University College of Medicine, Department of Paediatrics, USA 4 ATOM International, Newcastle upon Tyne, UK 5 Sarepta Therapeutics 6 UT Health, San Antonio, USA 7 WASHU, St Louis, USA 8 University of Iowa, USA

Goal of the study: To find out which physical therapy assessments (tests) are helpful for tracking changes in people with LGMD2A/R1

Why is this topic important to research?



Who took part in this research?

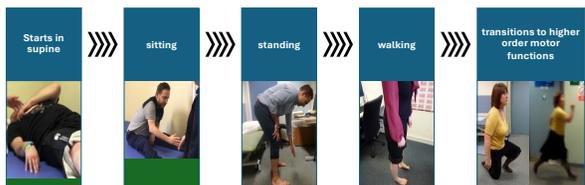
92 people with LGMD2A/R1. Ages ranged from 3 to 77 years old. Physical therapy tests took place at LGMD patient conferences in 2019 and 2023, Nationwide Children's Hospital and John Walton Muscular Dystrophy Research Centre



What tests were used?

North Star Assessment (NSAD)

29 items relevant to tasks of daily life- rolling over in bed/ getting out of bed/standing up/ picking up something off the floor/ stepping up a curb



100 meter timed test

This measures how fast someone can walk or run 100 meters. It shows overall mobility and speed.



Performance of upper limb

This test checks how well the arms and hands are working—from the shoulder to the fingers.



Lung function tests

These tests evaluate the muscles that help you breathe.



What did we learn?

The NSAD and PUL tests worked well for both people who can walk and those who use wheelchairs

100-meter timed test helped show how the disease affects walking speed.

LGMD2A/R1 affects people differently in the arms and legs. These tests measure both accurately

Most people have excellent hand and wrist function

A common test—getting up from the floor—was difficult for many people and not the best way to track changes

Lung function tests are helpful for tracking breathing abilities and helping with clinical care decisions

Summary: If you or your loved one has LGMDR1/2A, this research helps inform care teams around the world about using the most accurate and meaningful assessments to track the condition and plan treatment. It also brings us closer to developing effective treatments and trials, by making sure we're measuring the changes in strength and movement in the right way.

Acknowledgments: Thank you to Coalition to Cure Calpain 3 for funding support. The authors wish to acknowledge the time given and significant contributions of the patients and their families, and support of the LGMD International Patient Conference organisers.

This work has been supported by the NIHR Newcastle Biomedical Research Centre. The views expressed are those of the author(s) and not necessarily those of the NIHR or the Department of Health and Social Care.