

Outcome Study Fact Sheet

Study Name: **Rasch-analysis of Clinical Severity in FSHD (ROC-FSHD)**

Principal Investigator: Jeffrey Statland, MD
Assistant Professor, Department of Neurology
University of Kansas Medical Center. Kansas City, KS

Introduction and Purpose of the Study

Recent advances in our understanding of facioscapulohumeral muscular dystrophy (FSHD) have identified, for the first time since discovering the mutation behind FSHD 20 years ago, a potential target for therapy, and the research community has shifted towards clinical trial planning. However hampering these efforts are the wide variability in disease expression, which at its heart, may be due to the epigenetic nature of the disease. A barrier to identifying genetic or environmental modifiers of disease has been the lack of a clinically meaningful tool for documenting progression of disease. Such a rationally built scale would be calibrated so each increment in the scale would reflect a progression in the clinical disability of the disease. Such a scale would enable the identification of genetic and environmental modifiers of disease expression, while at the same time providing a powerful tool to stratify patients for future clinical trials, potentially reducing the variability and increasing the likelihood of identifying potentially effective therapeutics. In addition such a tool would be invaluable for prognosis and surveillance in the clinic. Here we propose to develop a scaled and calibrated Rasch-built clinical severity scale for FSHD (the FCSS).

How many people will participate?

We are seeking fifty (50) volunteers to participate in this research study, twenty-five (25) from the University of Kansas. Anyone with a diagnosis of FSHD who can travel to and from the University of Kansas Medical Center is eligible for this study.

If I am selected to come to KUMC, what will I do?

You will meet with Dr. Statland and his neuromuscular colleagues one time. Your visit will last approximately 6 hours, and can be split over two days. During the visit, you will have a physical examination, answer questions on how FSHD affects you, perform strength testing, and functional testing of your face, shoulders, and legs. We will also be collecting a blood and serum sample to help identify genetic modifiers and potential biomarkers for future studies.

How much will this cost me?

There is no cost to participate in this study.

Can you help me with my travel expenses?

We have limited funds to help with the cost of travel to and from the medical center and for parking.

Who can I call for more information?

For more information, please contact:

Ayla McCalley
Neuromuscular Research Center
University of Kansas Medical Center
3901 Rainbow Boulevard, Mailstop 2012
Kansas City, KS 66160
Phone: 913.945.9937
amccalley2@kumc.edu

THANK YOU FOR YOUR CONSIDERATION.