Title: Magnetic Resonance Imaging and Spectroscopy Biomarkers for Facioscapulohumeral Muscular Dystrophy

Sponsor: The FSH Society, Inc.

Study protocol number: NA_00065256

Principal Investigator: Kathryn R. Wagner, M.D., Ph.D.

Institution: The Hugo Moser Research Institute at Kennedy Krieger, Inc.

Purpose: This research study is being done to compare muscle imaging findings in people with facioscapulohumeral muscular dystrophy (FSHD) to people without FSHD. Both magnetic resonance imaging (MRI) and magnetic resonance spectroscopy (MRS) will be used to evaluate skeletal muscle in research participants. MRS is a type of imaging used to measure the levels of different chemicals in body tissues. It is done as part of a routine clinical MR exam in other parts of the body such as the brain, but has yet not been widely used in the musculoskeletal system. This research is being done to assess the effectiveness of MRS and MRI in helping doctors measure the type and severity of musculoskeletal disease. We also hope to use MRI and MRS in developing outcome measures that can be used in future clinical trials for FSHD.

Summary of protocol: The investigators will perform an MRI/MRS scanning protocol on the arm or leg muscles in a group of 30 adults with FSHD (confirmed through genetic testing). The MRI/MRS scanning protocol is non-invasive and will not require intravenous contrast injection. The duration of the scan will be between 1-2 hours.

The investigators will also perform MRI/MRS on groups of healthy volunteers and volunteers with other forms of muscle disease. These volunteers will be asked to undergo genetic testing for FSHD, which will include a single blood draw.

Participants who are enrolled in this study will be asked to visit the Kennedy Krieger Outpatient Center, where non-invasive strength testing will be done. This visit will also include timed function testing, where investigators will measure the amount of time it takes the participant to complete defined physical tasks (such as walking a distance of 30 feet). The MRI/MRS scan will be performed at the Johns Hopkins Hospital, located 2 blocks from the Kennedy Krieger Outpatient Center.

In addition to the protocol described, there are 2 optional sub-studies in which study volunteers can participate. The first sub-study involves performing a second MRI/MRS study on a different day from the first MRI/MRS study to assess how well these measurements can be repeated over time. In the second sub-study, the participants will have an MRS using a higher-power magnetic resonance imaging machine.

Inclusion criteria:
- 18 years of age or older
- For participants with FSHD: confirmed diagnosis of FSHD through genetic testing (participants will be asked to provide copies of genetic testing results)
- For participants without FSHD: a negative genetic test for FSHD or willingness to undergo genetic testing for FSHD

Exclusion criteria:
- Any contraindication to MRI scanning. This includes: metal fragments in the body, surgically implanted devices containing metal, severe claustrophobia, or inability to lie down in the MRI scanner for the duration of the study.
- History of scapular fixation surgery
- Pregnancy

Risks and benefits: The primary risks of participation in this study are associated with getting an MRI scan. Participants will be screened for contraindications to MRI scanning to minimize these risks. Other risks of the scan include bothersome noise levels and anxiety in enclosed spaces (claustrophobia).

Although there are no direct benefits to the participants in this study, the information gained in this study may be useful in future clinical trials in FSHD.

Compensation: At this time, the investigators are not offering compensation for participation in this research study.

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Resource links:

KKI website for directions to outpatient center

MRI website for JHH