FSHD Biomarker Study

Physicians and researchers at the University of Massachusetts Chan Medical School (UMass Chan) seek individuals with facioscapulohumeral muscular dystrophy (FSHD) to participate in an FSHD Biomarker Study. This will be conducted by Dr. Lawrence J. Hayward, M.D., Ph.D. This study focuses on explaining the variability of FSHD, especially within the same families, through examination of both genetics and other biomarkers.

Purpose:

The purpose of this study is to identify and understand genes that may explain why people with FSHD have different amounts of weakness in different muscles (different phenotypes). We also aim to identify biological markers and functional measurements that will enable us to follow and predict disease progression or indicate possible responses to treatment in upcoming FSHD clinical trials.

Participation:

Blood, saliva, muscle and/or skin samples from individuals with FSHD, some family members, and population controls are being accepted for this research study. Participants will be asked to complete a brief medical/family history questionnaire. Also, the clinicians will ask for permission to review the medical records of those with FSHD to understand the onset and progression of their disease.

The University of Massachusetts Chan Medical School will cover costs of the sample collection for participation, except for travel and housing. We are happy to help to make arrangements for the blood and saliva samples to be collected locally.

UMass Chan Wellstone Center for FSHD:

This study is a component of the Senator Paul D. Wellstone Cooperative Research Center for FSHD, sponsored by the National Institutes of Health. The overriding goal of the Center is to develop innovative therapies for FSHD. Research projects are conducted by an experienced team of collaborative investigators led by Charles P. Emerson, Ph.D. (UMass Chan), Louis Kunkel, Ph.D. (Children's Hospital of Boston), and Kathryn Wagner, M.D., Ph.D. (Kennedy Krieger Institute at Johns Hopkins School of Medicine). The Center also provides outreach to academic and industry partners and to patient advocacy groups such as the FSH Society to share research materials and to connect with individuals affected by FSHD.

Further information about the Center:
https://www.umassmed.edu/wellstone/

Benefits:

Although there are no direct benefits for those involved in this research, we are hopeful that understanding FSHD will lead to more effective screening, diagnosis, treatments, and ultimately a cure for this disease.

We greatly look forward to speaking with you to answer any questions you may have and to describe this study in more detail.

For more information, please contact:

Catherine Douthwright, PhD
Neurology Research Coordinator
Phone: 508-856-1524
catherine.douthwright@umassmed.edu

UMass Chan Medical School
55 Lake Ave. North
Worcester, MA 01655
Fax: (508) 856-4485

Requirements for participation:

To become involved, you must:

- Be diagnosed with Facioscapulohumeral muscular dystrophy (FSHD) or be a family member of someone with FSHD
- **OR** Be a control participant with no family history of FSHD
- Be willing to give a blood sample (approximately 8 teaspoons), or in some cases a saliva sample
- Be willing to consider giving a muscle and/or skin sample
- Be willing to complete questionnaires about your general medical/ family history
- Be willing to consider completing an MRI or muscle ultrasound
- Be willing to consider completing movement measurements

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University of Massachusetts Chan
Medical School
Department of Neurology
55 Lake Ave. North
Worcester, MA 01655
Phone: (508) 856-1524
Fax: (508) 856-4485
Catherine.Douthwright@umassmed.edu

FSHD Biomarker Study

at the

University of Massachusetts Chan Medical School

Study Docket #H000006581

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NIH National Institutes of Health