Genetic and epigenetic determinants of neuromuscular diseases
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We are an FSHD-focused research lab at the University of Nevada, Reno in the USA (https://med.unr.edu/jones-lab). We are conducting a new research project to improve FSHD diagnostics and to better understand the genetics and epigenetics of FSHD. We are currently enrolling participants from around the world.

Current genetic testing for FSHD, where available, is a complicated, expensive process that requires a visit to a healthcare professional for a blood draw and often a referral. We are investigating the utility of an alternative FSHD diagnostic approach that will be private, affordable and accessible to everyone worldwide.

We have developed an inexpensive technique capable of identifying epigenetic characteristics distinctly associated with FSHD1 or FSHD2, and not associated with healthy or other neuromuscular diseases, using DNA found in saliva. Here, we are recruiting participants to further validate this more affordable and accessible testing procedure to become an accepted and approved FSHD diagnostic option.

To date, this approach has been successful specifically identifying FSHD1 and FSHD2. Participants can choose to have their research results returned to them and will learn about their individual epigenetics and (some) genetics for their own FSHD region, and if they correlate with FSHD1, FSHD2, or neither. Those who do not wish to see their research results will nonetheless be helping develop this new technology for eventual diagnostic purposes.

This is a research project and not currently an approved medical or genetic diagnostic test. Your results, if you choose to have them returned to you, are confidential and will not become part of your medical record. Importantly, at this time, these research testing results will not meet inclusion criteria for clinical trials.

There is no cost to participate or to have the results of our research on your sample returned directly to you.

Who may want to participate? Anyone with a confirmed genetic diagnosis of FSHD, a clinical diagnosis of FSHD, a suspicion of FSHD, or at risk for FSHD by being in a family with FSHD may want to enroll. You do not have to have a diagnosis of FSHD or have prior genetic testing for FSHD. In addition, we are seeking healthy family members. Overall, anyone interested in learning about their FSHD-related epigenetic status may enroll in our research study.

Participation in this study is voluntary, private and confidential and is done in the privacy of your home. It is open to adults and children under 18, with written parental consent, anywhere in the world with access to mail.

Your privacy is paramount. We do NOT share your information. We do NOT share your DNA. We do NOT perform any additional genome sequence analysis without your express written consent. Results are confidential.

This project is supported from our general lab FSHD research budget at the University of Nevada, Reno, which includes funding from Friends of FSHD Research Foundation, MyFSHD, and direct donations from individuals interested in supporting this initiative to keep participation free and accessible to individuals around the world.

If you may be interested in participating in this FSHD research project or have any questions about this project, please contact us at peterjones@med.unr.edu and we will be happy to answer any questions by email or we can set up a phone or Skype call. We will send you a consent form for participation that will explain the procedure and potential risks and benefits. If everything is clear and you approve, we will arrange for a saliva collection kit(s) to be sent to your home for you and/or any family members who are interested and have provided written consent.

Please let us know if you have any questions or would like to participate and thank you for helping to make FSHD testing more affordable and accessible for the worldwide FSHD community.