

## ***News from The FSH Society, Inc.***

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## ***Press Statement***

### **FSH Society Hails First Study to Pinpoint Genetic Causes of Most Common Form of Muscular Dystrophy**

Watertown, MA--Daniel Perez, co-founder, President and CEO of the FSH Society and a 48-year-old patient with facioscapulohumeral muscular dystrophy (FSHD), the most common form of muscular dystrophy, hailed new findings,<sup>1</sup> published today in *Science* that revealed for the first time the biological mechanism causing FSHD. "This is a long-sought explanation of the exact biological workings of a disease that affects an estimated one in 14,000 or 22,100 Americans and 490,000 worldwide," he said, adding that this discovery "creates an enormous opportunity for research to develop ways to prevent or treat FSHD." The study, titled "A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy," was conducted by researchers from Leiden University in The Netherlands, the Fred Hutchinson Cancer Research Center in Seattle, Washington, and the University of Rochester Medical Center in New York.

"We are calling on the National Institutes of Health to immediately find ways to confirm and exploit these important findings, which I believe will generate great hope where there has been none," he said. "It is truly a potential watershed moment, and we are cautiously optimistic that we are entering into a new phase of research."

FSHD is a life-long, progressive, and severe loss of all skeletal muscles (face, shoulders, girdle and upper arms and progressing to the legs and feet). A crippling and life shortening disease, it is genetically and spontaneously (by mutation) transmitted to children, and it affects entire family constellations. Until now, there has been little definitive evidence of the exact genetic package that triggers the disease.

"These most recent findings begin to define a pathway to treatment for FSHD," said Professor David Housman of MIT, Chairman of the FSH Society's Scientific Advisory Board. "Understanding how a

quiescent ancestral gene that is seemingly tucked away in a forgotten corner of the human genome can reemerge to cause muscle weakness and wasting in tens of thousands of people around the world is a fascinating scientific story,” he said. “But more importantly, the knowledge that has emerged from tracking this complex story of DNA slipping and sliding into a deadly configuration opens the door to new ways to prevent damage from being done and an eventual return to health for victims of this very common form of muscular dystrophy.”

Said Nancy Van Zant, Executive Director, FSH Society: “Patients ask us every day if there is any hopeful research to share. We are always optimistic for them, but progress has seemed slow. Now, the reality that a specific genetic package of material leads to the toxicity causing the disease means that researchers can focus efforts to test medications on this defined target. Speaking as one of the leaders of this organization, I look forward to participating in the coordination of financial resources and patient efforts to help investigators carry this finding forward into treatments.”

## About the FSH Society

The FSH Society is a 501(c) (3) non-profit patient-led research, education and advocacy organization, founded in 1991. The Society frequently testifies before Congress on the research needs in FSHD. It has funded \$2.3 million in \$30,000-45,000 a year fellowships to more than five dozen researchers in 11 years, leading to well over a hundred publications in top tier journals.

Researcher(s) that published this *Science* paper have been recipients of FSH Society support and also serve as members of the FSH Society Scientific Advisory Board. Both the first and last authors, Drs. Richard Lemmers and Silvere van der Maarel began their careers in FSHD research as recipients of the prestigious FSH Society Marjorie Bronfman fellowship awards. The FSH Society has provided the Leiden team and many of their partners with funds to support early phases of their work.

The FSH Society is a small non-profit with a very dedicated and gifted group of scientific advisors, headed by Prof. David E. Housman, Massachusetts Institute of Technology, and along with its board of directors comprised mainly of patients and with the generous support of thousands of affected FSHD families and family foundations, the Society has helped to solve the mechanism causing FSHD. It has carefully raised funds and targeted many areas of research needed to be examined in FSHD and layered in the foundation for discovery.”

The FSH Society has funded many other projects with researchers around the globe who have published on FSHD in recent years; Drs. Jane Hewitt, Peter Jones, Ryan Wuebbles, Michael Kyba, Darko Bosnakovski, and Sara Winokur. All of these findings and papers along with the current breakthrough paper provide the context in which how to approach treating FSHD becomes apparent. This truly demonstrates the effectiveness of patient driven and disease focused organizations.

The FSH Society helps the DHHS NIH FSHD patient registry and existing DHHS NIH Sen. Paul D. Wellstone FSHD Cooperative Research Center at the Boston Biomedical Research Institute. The Society is composed of a board of directors of affected persons or family members and has over 4,000 members.

1. Lemmers *et al.* “A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy,” *Science*, published online August 19, 2010.

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