New test for FSHD muscular dystrophy sends diagnosis skyrocketing

Symptoms to be aware of this World FSHD Day


Zaharrah Age 10, FSHD muscular dystrophy

SYDNEY, NSW, AUSTRALIA, June 18, 2026 /EINPresswire.com/ -- Recently published research from the University of Nevada in the US and funded by FSHD Global Research Foundation in Sydney documents the innovations behind an at-home saliva test for one of the most common forms of muscular dystrophy—and incidence estimates are skyrocketing for what is being called 'the fastest growing genetic disease'.

FSHD (facioscapulohumeral dystrophy) is a muscle wasting disease that robs people of the ability to walk, talk, smile, blink or even eat

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> *Emma Watherley, CEO FSHD Global*

"FSHD is often misdiagnosed," says Emma Weatherley, CEO of FSHD Global Research Foundation who helped fund this research. "It is the fastest growing genetic disease as we improve access to testing. Many people who thought they were just clumsy or unathletic are being diagnosed properly now. Part of our efforts include educating the medical community as well as the wider public. There has been no government funding for this work, and that needs to change too."

Due to the complexity, expense, and general inaccessibility of FSHD genetic testing, many people with characteristic muscle weakness are never genetically confirmed, and at-risk relatives cannot get screened. This new test, currently for research use only, helps change that.

Dr Takako Jones, who leads the research at the University of Nevada, says, “This method improves sensitivity and helps broaden the definition of genetic FSHD to more accurately correspond to clinical FSHD, allowing identification of those at risk in affected families and in large population studies.”

Muscle weakness in FSHD typically develops asymmetrically, affecting only one arm or one leg, for example. Symptoms may appear years before a formal diagnosis by a doctor. For some people, the weakness may have developed so slowly that they were unaware they were experiencing muscle loss. Estimates of FSHD were once 1 in 20,000, but the numbers are climbing as research and testing improves. Current incidence of FSHD is estimated at 1:7500 people. That means over 3000 Australians are at risk.

“When individuals and families who may be affected sign up for our Cure FSHD patient registry,” says Weatherley, “we ask questions that help with FSHD research and allow us to determine if they should receive a free at-home saliva test kit and potentially further testing, such as our 3D MRI muscle scan. Whether or not patients choose to enroll in a clinical trial down the road, their participation in the registry is helping us understand FSHD in the population and advances research worldwide.”

FSHD Global Research Foundation has an impressive track record of supporting research, diagnostics, and advocacy, making incredible progress over the last 15 years, despite no government funding and little awareness in the medical community.

With World FSHD Awareness Day on Saturday, 20th June, the foundation can provide hope as well as awareness—it has already brought two clinical trials offering potential treatments for this complex form of muscular dystrophy to Australia, with more planned. But Australians wanting to participate need to get tested and their disease validated.

You can learn more about FSHD, the patient registry and testing, or offer your support by visiting the website: https://fshdglobal.org/

Link to FSHD saliva test research publication:
https://www.biorxiv.org/content/10.64898/2026.05.26.727947v1?

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