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Researchers identify gene associated with muscular dystrophy-related vision problems

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Skeletal muscle disease and vision deficits might seem unrelated, but a frog model of muscular dystrophy shows it is not such a leap.

Facioscapulohumeral muscular dystrophy, or FSHD, is the world's third most common type of muscular dystrophy. It is characterized by progressive skeletal muscle weakening in the face, shoulders, and upper arms. Over half of FSHD patients (also known as Landouzy-Dejerine syndrome) also have abnormal blood vessels in the back of the eye, which can cause vision problems.

Over 95% of FSHD patients carry a genetic abnormality proposed to affect expression of the FRG1 gene, and previous studies of FRG1 in frogs demonstrate that it is important for skeletal muscle development. Therefore, University of Illinois scientists investigated the possibility that the FRG1 gene might also be responsible for the blood vessel abnormalities in FSHD patients' eyes. Their report published in Disease Models & Mechanisms (DMM), describes how they examined the FRG1 gene in the frog and found the protein that it encodes for is highly expressed in blood vessels. Additional experiments show that normal FRG1 protein expression is important for blood vessel growth and organization.

Previous work linked aberrant FRG1 expression to the skeletal muscle defects of FSHD, and this study shows that FRG1 expression similarly contributes to abnormal blood vessel growth in the retina. Thus, the FSHD mutation, by affecting expression of the FRG1 gene may

contribute to both the skeletal muscle and visual deficits associated with FSHD pathology. This study supports the idea that FRG1 expression is a main cause of this type of muscular dystrophy and might be an important target for therapeutic intervention.

Source: The Company of Biologists (news : web)