January 4, 2012
FSH Society

Breaking News

This new research published in *Developmental Cell* identifies a set of genes regulated by DUX4 and shows them to be aberrantly expressed in FSHD muscle. The set of genes regulated by DUX4 suggest possible mechanisms for causing muscle damage that might be targeted for developing therapies, and also might be good biomarkers to measure the response to future therapies. To read or obtain more information on the article please visit *Developmental Cell* @ http://www.cell.com/developmental-cell/abstract/S1534-5807%2811%2900523-5?switch=standard

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Dev Cell. 2011 Dec 28. [Epub ahead of print]

DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy

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Abstract
Facioscapulohumeral dystrophy (FSHD) is one of the most common inherited muscular dystrophies. The causative gene remains controversial and the mechanism of pathophysiology unknown. Here we identify genes associated with germline and early stem cell development as targets of the DUX4 transcription factor, a leading candidate gene for FSHD. The genes regulated by DUX4 are reliably detected in FSHD muscle but not in controls, providing direct support for the model that misexpression of DUX4 is a causal factor for FSHD. Additionally, we show that DUX4 binds and activates LTR elements from a class of MaLR endogenous primate retrotransposons and suppresses the innate immune response to viral infection, at least in part through the activation of DEFB103, a human defensin that can inhibit muscle differentiation. These findings suggest specific mechanisms of FSHD pathology and identify candidate biomarkers for disease diagnosis and progression.
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Source

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Abstract

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PMID:
22209328
[PubMed - as supplied by publisher]

http://www.ncbi.nlm.nih.gov/pubmed/22209328

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