

**Title:** Dux4 target gene expression in mouse muscle transplanted with muscle cells from FSHD patients

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**Abstract:** Facioscapulohumeral Muscular Dystrophy (FSHD) is one of the most prevalent forms of muscular dystrophy. However, because of the unique nature of the genetic abnormality underlying the disease, there is currently no widely available laboratory model. In order to gain insights into FSHD molecular pathology, we developed a xenograft model by transplanting myogenic cells from patients with FSHD (4qA contractions) as well as from their unaffected relatives into the tibialis anterior muscles of immunodeficient mice. Our findings show that muscle xenografts derived from FSHD myogenic cells express Dux4 target genes, recapitulating the expression of these disease biomarkers in muscle biopsies of FSHD patients. FSHD muscle xenografts provide an animal model for investigations of the molecular pathogenesis of FSHD muscles and for drug development.