



THERAPEUTIC, TARGET

Therapeutic targets to treat Facioscapulohumeral Dystrophy [FSHD]

Brief Technology Description

Methods to treat Facioscapulohumeral Dystrophy [FSHD] by modulating chromatin repression

BUSINESS OPPORTUNITY

Exclusive licence
Non-exclusive license
Sponsored research

TECHNOLOGY TYPE

Therapeutic
Target

STAGE OF DEVELOPMENT

Preclinical *in vitro*

PATENT INFORMATION

[PCT/US2019/016315](https://patents.google.com/patent/PCT/US2019/016315)

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Tech ID: 17-002
partnering@fredhutch.org
206-667-4304

Technology Overview

Facioscapulohumeral muscular dystrophy [FSHD] is a disease with no currently approved treatments characterized by progressive muscle weakness and atrophy. FSHD is caused by ectopic expression of Double Homeobox 4 [DUX4], a gene normally only expressed early in development. In somatic tissue, DUX4 expression is epigenetically silenced through the presence of repeated sections of DNA called D4Z4 arrays. Mutations in the number of D4Z4 arrays or in chromatin repressors of D4Z4 underlie the molecular etiology of FSHD. Fred Hutchinson researchers have demonstrated in both skeletal muscle and induced pluripotent stem cells that the Nucleosome Remodeling Deacetylase [NuRD] and chromatin assembly factor-1 [CAF-1] complexes are necessary components which repress DUX4 expression by binding to D4Z4 arrays. In addition, they show that DUX4 presence mediates Methyl-CpG-binding domain protein 3-like [MBD3L] factors, which reduce D4Z4-mediated DUX4 repression. Taken together, these findings identify specific complexes which modulate DUX4 expression and reveal novel therapeutic targets to treat FSHD.

Applications

- Drug Discovery Efforts: Agonism or NuRD and/or CAF-1 complexes, Inhibition of MBD3L proteins
- Therapeutic treatments for FSHD
- Transplantation of engineered muscle cells to treat FSHD

Potential Advantages

- Reveals for the molecular basis for DUX4 amplification in FSHD muscle cells

Market Overview: FSHD occurs in the population with an estimated prevalence of 1 in 20,000 people. Currently, there are no approved treatments on the market, demonstrating a need for novel therapeutics.

Investigator Overview

Stephen Tapscott, MD, PhD, Human Biology and Clinical Research Divisions