AGENDA

Day 1: Thursday, June 24
(All times reflect Eastern Daylight Savings Time)

10:00 AM
Welcoming Remarks

10:07 AM
Keynote: The history of FSHD in a large Utah kindred: The fruits of 80+ years of engagement
Russel Butterfield, M.D., Ph.D., University of Utah

10:43 AM
Session 1: Discovery Research
Session Chairs: Lawrence Hayward, M.D., Ph.D. / Kyoko Yokomori, Ph.D.

Transient DUX4 expression provokes long-lasting cellular and molecular muscle alterations
Darko Bosnakovski, DVM, Ph.D., University of Minnesota

Identification of the first endogenous inhibitor of DUX4 in FSHD muscular dystrophy
Paola Ghezzi, M.S., San Raffaele Scientific Institute

Use of snRNA-seq to characterize the skeletal muscle microenvironment during pathogenesis in FSHD
Anugraha Raman, Fulcrum Therapeutics

11:35 AM
Session 2: Genetics & Epigenetics
Session Chairs: Kyoko Yokomori, Ph.D. / Jessica C. de Greef, Ph.D.

Identification of a druggable epigenetic target required for DUX4 expression and DUX4-mediated toxicity in FSHD muscular dystrophy
Emmanuel Mocciaro, Ph.D., San Raffaele Scientific Institute

Accessing D4Z4 (epi)genetics with long-read sequencing
Quentin Gouil, M.D., Walter and Eliza Hall Institute of Medical Research

12:09 PM
Break

12:24 PM
Session 3: Pathology & Disease Mechanisms
Session Chairs: Sujatha Jagannathan, Ph.D. / Davide Gabellini, Ph.D.

System biology approach links muscle weakening to alteration of the contractile apparatus in FSHD
Camille Laberthonnière, M.S., Marseille Medical Genetics

Targeting DUX4 Post-translational Modifications in vitro Protects Against DUX4-Mediated Toxicity
Renatta Knox M.D., Ph.D., Nationwide Children’s Hospital

Genetic engineering and characterization of isogenic FSHD mutant myocytes
Nam Nguyen, M.S., University of California, Irvine

IL-6 and TNFα are key inflammatory cytokines in facioscapulohumeral muscular dystrophy
Anna Greco, M.D., M.S., Radboud University Medical Center

1:33 PM
Special Session – Fulcrum’s Phase 2b ReDUX Results
A Phase 2, Randomized, Double-Blind, Placebo-Controlled, 48-Week, Parallel-Group
Session Chair: Karlien Mul, M.D., Ph.D.
Rabi Tawil, M.D., University of Rochester Medical Center
Study of the Efficacy and Safety of Losmapimod in Treating Subjects with Facioscapulohumeral Muscular Dystrophy (FSHD) with Open Label Extension (OLE): ReDUX4

1:58 PM
Fulcrum Panel Q&A
Moderator: Michelle Mellion, M.D., Fulcrum Therapeutics

2:15 PM
Day 1 Closing remarks

Day 2: Friday, June 25
(All times reflect Eastern Daylight Savings Time)

10:00 AM
Welcoming Remarks
Stephen Tapscott, M.D., Ph.D., Fred Hutchinson Cancer Research Center

10:03 AM
Keynote: Aspects of immune response and FSHD
Session Chairs: Jessica C. de Greef, Ph.D. / Kyoko Yokomori, Ph.D.

10:44 AM
Session 4: Biomarkers
Identifying biomarkers for facioscapulohumeral muscular dystrophy using Olink Proteomics
Amy Campbell, Ph.D., University of Colorado Anschutz Medical Campus
SLC34A2 as a Protein Biomarker of FSHD
Robert Bloch, Ph.D., University of Maryland School of Medicine
Serum Interluekine-6 levels as severity biomarker in FSHD1
Jonathan Pini, Ph.D., Université Côte d’Azur

11:08 AM
Biomarker Panel Discussion
Moderators: Jessica C. de Greef, Ph.D. / Kyoko Yokomori, Ph.D.

11:24 AM
Session 5: Interventional Strategies
Session Chairs: Davide Gabellini, Ph.D. / Lawrence Hayward, M.D., Ph.D.
Persistence of p38-independent DUX4 target gene expression in FSHD xenografts
Fran Sverdrup, Ph.D., Saint Louis University
Human miRNA mir-675 inhibits DUX4 expression and may be exploited as a potential treatment for Facioscapulohumeral muscular dystrophy
Nizar Saad, Ph.D., Nationwide Children’s Hospital

11:54 AM
Break

12:06 PM
Session 6: Antisense Strategies
Session Chair: Scott Harper, Ph.D.
Systemic delivery of a DUX4 targeting antisense oligonucleotide reduces DUX4, DUX4 responsive genes, and pathology in skeletal muscles of ACTA1-MCM; FLExDUX4 mice
Linde Bouwman, M.S., Leiden University Medical Center
DUX4 siRNA Optimization for the Development of an Antibody-Oligonucleotide Conjugate (AOCTM) for the Treatment of FSHD
Barbora Malecova, Ph.D., Avidity Biosciences
Lipid-conjugated DUX4-targeting siRNA therapeutic to treat FSHD
Katelyn Daman, Ph.D., University of Massachusetts Medical School
FORCE Platform Enables Muscle Targeted Delivery of Antisense Oligonucleotide and Silencing DUX4 Activity in an FSHD Patient Cell Line
Nelson Hsia, Ph.D., Dyne Therapeutics
Systemic antisense therapeutics inhibiting DUX4 expression ameliorates FSHD-like pathology in an FSHD mouse model

Development of an RNAi Therapeutic, ARO-DUX4 for the Treatment of FSHD

Translating DUX4-targeted RNAi-based Gene Therapy for FSHD

**Antisense Strategies Panel Discussion**

**Session 7: Clinical Studies & Outcome Measures**

Five year follow-up study on quantitative MRI in facioscapulohumeral muscular dystrophy

Objective Monitoring of Facioscapulohumeral Dystrophy During Clinical Trials using a Smartphone Application and Wearables

FSHD European Trial Network

**Best Poster Prize & Young Scientist Award**

**2022 IRC Announcement & final remarks**

Adjourn