28th Annual FSHD Society

International Research Congress

JUNE 24-25, 2021

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**	**Session Chair:** Karlien Mul, M.D., Ph.D.
	A Phase 2, Randomized, Double-Blind, Placebo-Controlled, 48-Week, Parallel-Group	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**	
10:03 AM	**Keynote: Aspects of immune response and FSHD**	Stephen Tapscott, M.D., Ph.D., Fred Hutchinson Cancer Research Center
10:44 AM	**Session 4: Biomarkers**	**Session Chairs:** Jessica C. de Greef, Ph.D. / Kyoko Yokomori, Ph.D.
	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	Ngoc Lu-Nguyen, M.D., Royal Holloway University of London
	Development of an RNAi Therapeutic, ARO-DUX4 for the Treatment of FSHD	Jonathan Van Dyke, Ph.D., Arrowhead Pharmaceuticals
	Translating DUX4-targeted RNAi-based Gene Therapy for FSHD	Lindsay Wallace, M.D., Nationwide Children's Hospital
	Antisense Strategies Panel Discussion	**Moderator:** Scott Harper, Ph.D.
1:23 PM	**Session 7: Clinical Studies & Outcome Measures**	**Session Chairs:** Karlien Mul, M.D., Ph.D. / Sabrina Sacconi, Ph.D.
	Five year follow-up study on quantitative MRI in facioscapulohumeral muscular dystrophy	Sanne Vincenten, M.S., Radboud University Medical Center
	Objective Monitoring of Facioscapulohumeral Dystrophy During Clinical Trials using a Smartphone Application and Wearables	Ghobad Maleki, M.S., Centre for Human Drug Research
	FSHD European Trial Network	Nicol Voermans, M.D., Ph.D., Radboud University Medical Center
2:04 PM	**Best Poster Prize & Young Scientist Award**	**Moderator:** Sujatha Jagannathan, Ph.D.
2:24 PM	**2022 IRC Announcement & final remarks**	
2:30 PM	Adjourn	