31st Annual FSHD Society

International Research Congress

Where the world's leading clinicians, scientists, companies, and advocates gather to accelerate research toward treatments and a cure

June 13-14, 2024 • Hilton Denver City Center • Denver, Colorado

FSHD SOCIETY
LIGHTING THE WAY TO A CURE

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ABSTRACTS

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WELCOME TO THE MILE HIGH CITY

Welcome to Denver, Colorado, and the 31st Annual International Research Congress on FSHD, organized and hosted by the FSHD Society. We hope you have time to enjoy the natural beauty of the mountains of Colorado during your visit.

This year, clinicians and researchers from around the globe will gather to discuss the advances that have been made in our scientific understanding of FSHD and the devastating effects of the toxic mis-expression of *DUX4* in human skeletal muscle. Significant focus will be placed on our growing understanding of pediatric disease as MRI imaging continues to reveal the nature of FSHD in youth and adults alike. We will discuss further development of clinical outcome measurements as we work to engage regulators and payers on the details of the disease and how its impact on human health can be accurately measured in the clinic.

Significant time will be spent discussing clinical trials and recent therapeutic developments in FSHD. New findings will be presented in ongoing clinical trials in FSHD, and plans for future studies will be discussed as we continue to seek meaningful therapeutic benefit from 31 years of intense work on this devastating disease.

KEYNOTE SPEAKERS

JEFF JOHNSTON received an FSHD diagnosis when he was 31 years old. Now at 54, he has lost the ability to walk and much of his upper body strength, rendering him unable to live independently. He experiences chronic pain and regularly modifies his daily routines to manage the progressive muscle atrophy. Despite these challenges, Jeff continues to work full-time as an economist at a commercial bank. He is married and has two daughters who are 19 and 22 years old.

SILVÈRE VAN DER MAAREL, PhD, is a professor of medical epigenetics and chair of the Department of Human Genetics in Leiden (LUMC, the Netherlands). He was trained as a human geneticist at the Radboud University Nijmegen Medical Center in the Netherlands, where he was involved in the positional cloning of X-linked disease genes. Prof. van der Maarel's scientific interests focus on the genetic and epigenetic regulation of repetitive DNA in the human genome in relation to disease. His main research interest is facioscapulohumeral muscular dystrophy (FSHD), a muscle disease caused by genetic and epigenetic changes in a repetitive DNA structure on the tip of the long arm of chromosome 4. The unprecedented genetic mechanism of FSHD has challenged scientists for many years. Reaching a consensus about this mechanism was a critical step toward current therapy developments, some of which are now in clinical trials. However, maintaining basic research on its underlying pathology will be necessary to fully understand this disease and to address the many intriguing observations for which we currently lack explanations.

RICHARD FINKEL, MD, holds the George J. Pedersen Endowed Chair in Neurotherapeutics at St. Jude Children's Research Hospital in Memphis, Tennessee, where he serves as director of the Center for Experimental Neurotherapeutics. After earning his MD at Washington University in St. Louis, Dr. Finkel trained as a neurology fellow at Harvard Medical School and Children's, Beth Israel, Brigham and Women's, and VA hospitals in Boston. His internship and residency in pediatrics were at Children's Hospital Boston. Dr. Finkel has developed outcome measures, biomarkers, and clinical trial design for pediatric neurologic diseases, and has participated in clinical trials with investigational drugs for pediatric neurologic and metabolic diseases. His work has led to understanding the trajectories of change in spinal muscular atrophy, Duchenne muscular dystrophy, and Charcot-Marie-Tooth inherited neuropathies.

PROGRAM COMMITTEE

Lucienne Ronco, PhD (organizing chair)
Sabrina Sacconi, MD PhD (co-chair)
Jeffrey Statland, MD (co-chair)
Katy de Valle, PT
Scott Harper, PhD
Sujatha Jagannathan, PhD
Renatta Knox, MD PhD
Lawrence Korngut, MD
Richard Lemmers, PhD
Linda Lowes, PT PhD
Ian Woodcock, MD

SLIDE PRESENTATIONS—DAY 1

Thursday, June 13, 2024 • Colorado Ballroom F–J

8:00–9:00 a.m.
REGISTRATION & BREAKFAST BUFFET

9:00 a.m.
WELCOME:
Lucienne Ronco, *Organizing Chair,* & Jeffrey Statland, *Co-chair*

9:05 a.m.
KEYNOTE: Stripped away: The real impact of FSHD on daily life
Jeff Johnston, *patient advocate*

9:20 a.m.
KEYNOTE: FSHD – A genetic odyssey
Silvère van der Maarel, *Leiden University Medical Center*

10:05 a.m.–1:00 p.m.
SESSION 1: Disease Mechanisms & Interventional Strategies
Session Chairs: Scott Harper & Renatta Knox

10:05 a.m.
S1.01 Engineered FSHD mutations result in D4Z4 heterochromatin disruption and feedforward *DUX4* network activation
Kyoko Yokomori, *University of California, Irvine*

10:25 a.m.
S1.02 Identification of novel druggable activator of *DUX4* expression in FSHD muscular dystrophy
Emanuele Mocciaro, *San Raffaele Scientific Institute*

10:45 a.m.
S1.03 Understanding and treating inflammation in FSHD muscular dystrophy
Beatrice Biferali, *San Raffaele Scientific Institute*

11:05–11:20 a.m.
COFFEE BREAK & NETWORKING
Lower level 2 foyer

11:20 a.m.
S1.04 Creating an immune cell atlas of the peripheral blood for facioscapulohumeral muscular dystrophy (FSHD)
Chantal Coles, *Murdoch Children's Research Institute*

11:40 a.m.
S1.05 Establishing a clinical candidate for FSHD RNAi gene therapy
Lindsay Wallace, *Nationwide Children's Hospital*

12:00 p.m.
S1.06 Developing a potent gene therapy candidate for facioscapulohumeral muscular dystrophy (FSHD) through high-throughput AAV capsid and cargo engineering
Sharif Tabebordbar, *Kate Therapeutics*

12:20 p.m.
S1.07 Efficacy and safety of an investigational single-dose epigenome editing therapy, EPI-321, targeting D4Z4 in facioscapulohumeral muscular dystrophy
Alexandra Collin de l'Hortet, *Epic Bio*

12:40 p.m.
S1.08 Translating AAV-delivered CRISPR-Cas13 therapy for FSHD may require overcoming size and immune challenges
Manal Ali, *Abigail Wexner Research Institute, Nationwide Children's Hospital*

1:00–2:00 p.m.
LUNCH BREAK & POSTER VIEWING
Colorado Ballroom A-E

2:00–3:00 p.m.
SESSION 2: Preclinical Models
Session Chairs: Scott Harper & Renatta Knox

2:00 p.m.
S2.01 Mstn is a reliable biomarker for monitoring therapy effect in FSHD
Julie Dumonceaux, *NIHR Biomedical Research Centre, University College London*

2:20 p.m.
S2.02 *SLC34A2:* A biomarker for FSHD in muscle *in situ*
Robert Bloch, *University of Maryland School of Medicine*

2:40 p.m.
S2.03 The FORCE™ platform achieves robust and durable *DUX4* suppression and functional benefit in FSHD mouse models
Thomas Natoli, *Dyne Therapeutics*

3:00–5:20 p.m.
SESSION 3: Outcomes Assessments
Session Chairs: Jeffrey Statland & Linda Lowes

3:00 p.m.
S3.01 Strength and functional correlates of reachable workspace in facioscapulohumeral muscular dystrophy
Leo Wang, *University of Washington*

SLIDE PRESENTATIONS—DAY 1

Thursday, June 13, 2024 • Colorado Ballroom F–J

3:20 p.m.
S3.02 A 5-year natural history cohort of patients with facioscapulohumeral muscular dystrophy determining disease progression and feasibility of clinical outcome assessments for clinical trials
Joost Kools, *Radboud University Medical Center*

3:40–4:00 p.m.
COFFEE BREAK & NETWORKING
Lower level 2 foyer

4:00 p.m.
S3.03 Exploring individual muscle progression and *DUX4* associations in FSHD
Seth Friedman, *Seattle Children's Hospital*

4:20 p.m.
S3.04 Interleukin-6 as a biomarker for disease activity, progression, and muscle composition in facioscapulo-humeral dystrophy: Insights from longitudinal studies
Jonathan Pini, *University Côte d'Azur, INSERM, CNRS, IRCAN*

4:40 p.m.
S3.05 Developing a cell-free DNA-based biomarker for FSHD
Sujatha Jagannathan, *University of Colorado Anschutz Medical Campus*

5:00 p.m.
S4.05 *DUX4* double whammy: The transcription factor that causes a rare muscular dystrophy also kills the precursors of the human nose
Kaoru Inoue, *National Institute of Environmental Health Science/National Institutes of Health*

5:20 p.m.
CLOSING REMARKS & BRIEF BREAK

5:30–8:30 p.m.
RECEPTION & POSTER SESSION
Colorado Ballroom F–J

8:30 p.m.
ADJOURN

SLIDE PRESENTATIONS—DAY 2

Friday, June 14, 2024 • Colorado Ballroom F–J

8:00–9:00 a.m.
REGISTRATION & BREAKFAST BUFFET

9:00 a.m.
WELCOME:
Lucienne Ronco, *Organizing Chair,* & Sabrina Sacconi, *Co-chair*

9:05 a.m.
KEYNOTE: Deconstructing gene-directed therapies for neuromuscular diseases
Richard Finkel, *St. Jude Children's Hospital*

9:50–11:30 a.m.
SESSION 4: Genetics & Discovery Research
Session Chairs: Sujatha Jagannathan & Richard Lemmers

9:50 a.m.
S4.01 Single-cell spatial transcriptomics reveals a dystrophic trajectory following a developmental bifurcation of FSHD myoblast cell fates
Xiangduo Kong, *University of California, Irvine*

10:10 a.m.
S4.02 Clinical variability in FSHD: The importance of robust clinical information for reliable interpretation of genetic data
Richard Lemmers, *Leiden University Medical Center*

10:30 a.m.
S4.03 Enhancing FSHD diagnosis: A 1-year follow-up study for validating the methylation assay in the clinical practice
Claudia Strafella, *Genomic Medicine Laboratory-UILDM, Santa Lucia Foundation IRCCS*

10:50–11:10 a.m.
COFFEE BREAK & NETWORKING
Lower level 2 foyer

11:10 a.m.
S4.04 Modeling cell type-specific and sporadic *DUX4* gene expression in FSHD
Mitsuru Sasaki-Honda, *IBBTEC, University of Cantabria/ CiRA, Kyoto University*

SLIDE PRESENTATIONS—DAY 2

Friday, June 14, 2024 • Colorado Ballroom F–J

11:30 a.m.–12:30 p.m.
SESSION 5: Pediatric FSHD
Session Chairs: Ian Woodcock & Linda Lowes

11:30 a.m.
S5.01 Longitudinal insights into childhood-onset facioscapulohumeral dystrophy: A 5-year natural history study
Jildou Dijkstra, *Radboud University Medical Center*

11:50 a.m.
S5.02 Pediatric FSHD in the US: Results from the US National Registry for FSHD
Natalie Katz, *Duke University*

12:10 p.m.
S5.03 Longitudinal outcomes in pediatric FSHD: An Australian cohort study
Ian Woodcock, *The Royal Children's Hospital, Melbourne*

12:30–1:50 p.m.
LUNCH BREAK & POSTERS
Colorado Ballroom A-E

1:50–5:10 p.m.
SESSION 6: Clinical Studies & Trial Designs
Session Chairs: Sabrina Sacconi & Lawrence Korngut

1:50 p.m.
S6.01 Pain impacts quality of life, psychological disorders, and exercise in a large international cohort of patients with facioscapulohumeral muscular dystrophy
Renatta Knox, *Washington University School of Medicine*

2:10 p.m.
S6.02 Motor Outcomes to Validate Evaluations in Facioscapulohumeral Muscular Dystrophy (MOVE FSHD): Interim baseline data and potential predictors for FSHD
Michaela Walker, *University of Kansas Medical Center*

2:30 p.m.
S6.03 FSHD Global Registry Project: Whole-body MRI muscle-level analysis for advancing research and empowering patients
Silvia Blemker, *University of Virginia/Springbok Analytics* and Emma Weatherly, *FSHD Global Research Foundation*

2:50 p.m.
S6.04 Analyzing phenotypes in FSHD: An update of the Comprehensive Clinical Evaluation Form
Giulia Ricci, *Department of Clinical and Experimental Medicine, University of Pisa*

3:10 p.m.
S6.05 Reassessing clinical phenotypes in facioscapulohumeral muscular dystrophy: Late-onset FSHD presentations
Sabrina Sacconi, *University Côte d'Azur, CHU Nice, France*

3:30–3:50 p.m.
COFFEE BREAK & NETWORKING
Lower level 2 foyer

3:50 p.m.
S6.06 FSHD disease progression and losmapimod efficacy assessed by reachable workspace in both arms
Joost Kools, *Radboud University Medical Center*

4:10 p.m.
S6.07 Safety and tolerability study of clenbuterol in facioscapulohumeral muscular dystrophy
Rebecca Clay, *University of Kansas Medical Center*

4:30 p.m.
S6.08 ReInForce: A bicentric, randomized, double-blind, placebo-controlled pilot study to evaluate the efficacy and safety of satralizumab in FSHD1
Jonathan Pini, *University Côte d'Azur, INSERM, CNRS, IRCAN*

4:50 p.m.
S6.09 Interim results from FORTITUDE™, a randomized Phase 1/2 trial evaluating AOC 1020 in adults with FSHD
Jeffrey Statland, *University of Kansas Medical Center*

5:10 p.m.
Best Poster Award & Young Investigator Award
Conference program committee

5:20 p.m.
Closing remarks
Sabrina Sacconi & Lucienne Ronco

5:25–6:00 p.m.
Adjourn & break

6:00–8:00 p.m.
Joint IRC & FSHD Connect Reception
The Plaza, hotel main level outdoors

POSTER PRESENTATIONS

Colorado Ballroom A–E

The main poster session is on Thursday, June 13, 5:30-8:30 p.m. Please plan to present your poster on the following schedule: Odd-numbered posters from 5:30-7:00 p.m. Even-numbered posters from 7:00-8:30 p.m. The poster hall will also be open at lunch on both days of the meeting.

DISEASE MECHANISMS & INTERVENTIONAL STRATEGIES

P1.01 **A novel proviral plasmid reduces cross-packaging and ITR promoter activity in AAV vector preparations**
Pranali Mistry, Noah Taylor, Scott Harper, Oliver King, Matthew Guggenbiller

P1.02 **Anti-fibrotic approach ameliorates muscle pathology in FSHD animal model**
Haseeb Ahsan, Ana Mitanoska, Kenric Chen, David Oyler, Michael Kyba, Darko Bosnakovski

P1.03 **Characterization of the *DUX4*-MATR3 complex to design a possible therapy for FSHD**
Chiara Zucchelli, Andrea Berardi, Giacomo Quilici, Valeria Runfola, Maria Pannese, Paola Ghezzi, **Davide Gabellini,** Giovanna Musco

P1.04 **Deficits in the unfolded protein response in FSHD myoblasts**
Adam Bittel, Yi-Wen Chen

P1.05 ***DUX4*-associated hypoxia signaling impairs oxidative metabolism and shifts towards less oxidative muscle fibers**
Justin Cohen, Vincent Ho, Keryn Woodman, Angela Lek, Alec DeSimone, Monkol Lek

P1.06 ***DUX4* level-dependent sarcolemmal repair deficits in FSHD**
Adam Bittel, Aiping Zhang, Ze Chen Yi-Wen Chen

P1.07 **Fibro-adipogenic progenitors and FSHD**
Carlo Serra, Kathryn Wagner, Andrew Wilson, Thomas Lloyd

P1.08 **Nucleolar stress, apoptosis, and FSHD myopathy**
Carlo Serra, Kathryn Wagner, Andrew Wilson, Thomas Lloyd

P1.09 **Scapulothoracic fusion using high-strength suture tape cerclage for the treatment of facioscapulohumeral muscular dystrophy**
Michael McDermott, Neil Lancaster, Joey Bell, Brandon Merryman, Anthony Romeo

P1.10 **Selection of peptides for a muscle-targeted delivery of ASOs directed against *DUX4* mRNAs through complementary approaches *in silico*, *in vitro,* and *in vivo***
Maëlle Limpens, Aline Derenne, Carmen Burtea, Sophie Laurent, Alexandre Legrand, Steve Wilton, Anne-Emilie Declèves, Alexandra Belayew, Frédérique Coppée, Alexandra Tassin

P1.11 **Short- and long-term systemic treatment of the ACTA1-MCM/FLExDUX4 mice with an AAV-sh*DUX4***
Julie Dumonceaux, Solene Sohn, Sophie Reid, Maximilien Bowen, Emilio Corbex, Baptiste Morel-Prieur, Christophe Hourde, Virginie Mariot

P1.12 **The prevalence of sleep disorders among individuals with facioscapulohumeral dystrophy: A review of the literature**
Nedra Whitehead, Joyce Alese, Michael Enger, Christine Hill, Jamie Zimmerman, Barbara Do, Amy Moore, Shiny Thomas, Julie Royer, Swamy Venkatash, James Howard

P1.13 **The roles of flavones and autophagy in *DUX4* toxicity**
Kristen Woods

P1.14 **Unraveling the role of non-myogenic mesenchymal cells in FSHD pathogenesis and investigating the impact of human amniotic stromal cell conditioned medium on non-myogenic mesenchymal cell functionality**
Lorena Di Pietro, Andrea Papait, Flavia Giacalone, Alessandra Nagar, Gaia Guardabascio, Elvira Ragozzino, Antonietta Silini, Pietro Romele, Sara Bortolani, Eleonora Torchia, Enzo Ricci, Ornella Parolini

POSTER PRESENTATIONS

Colorado Ballroom A–E

PRECLINICAL MODELS

P2.01 **A comprehensive functional force velocity endurance (FoVE) model reveals unsuspected muscle properties affected by the expression of *DUX4***
Julie Dumonceaux, Maximilien Bowen, Solene Sohn, Virginie Mariot, Baptiste Morel-Prieur, Christophe Hourde

OUTCOMES ASSESSMENTS

P3.01 **A comprehensive analytical description of atypical features in FSHD1 patients from the French FSHD registry**
Benoît Sanson, Abderhmane Slioui, Caroline Stalens, Lori Klouvi, Céline Guien, Hadrien Delattre, Sitraka Rabarimeriarijaona, Rafaëlle Bernard, Christophe Béroud, Sabrina Sacconi, the French FSHD Registry Collaboration Group

P3.02 **Cohort profile: Clinical characteristics of patients with facioscapulohumeral muscular dystrophy from Russian Registry**
Aysylu Murtazina, Anna Kuchina, Darya Sherstyukova, Nikolay Zernov, Artem Borovikov, Inna Sharkova, Elena Dadali, Mikhail Skoblov

P3.03 **Enhancing clinical trial eligibility criteria in FSHD: Validating whole-body MRI as a key outcome measure**
Per Widholm, Markus Karlsson, Jonathan Pini, Guillaume Bassez, Benjamin Marty, Teresinha Evangelista, Romain Thomas, Loïc Danjoux, Céline Tard, Sabrina Sacconi

P3.04 **Patterns of fat distribution from individual muscles across whole-body MRI (iWBMRI): Visualizing and quantifying features**
Lara Riem, Olivia DuCharme, Xue Feng, Allison Kenney, Jacob Morris, Doris Leung, Seth Friedman, Emma Weatherley, Silvia Blemker

P3.05 **The Dutch Registry for Facioscapulohumeral Muscular Dystrophy: Cohort profile and longitudinal patient-reported outcomes**
Joost Kools, Hanneke Deenen, Nicol Voermans

P3.06 **The facioscapulohumeral muscular dystrophy Rasch-built Overall Disability Scale (FSHD-RODS): Longitudinal assessment of a disease-specific patient-reported outcome measure in a clinical trial timeframe**
Sjan Teeselink, Sanne Vincenten, Nicol Voermans, Baziel van Engelen, Karlien Mul

GENETICS & DISCOVERY RESEARCH

P4.01 **4qA D4Z4 methylation test as a valuable complement for differential diagnosis in patients with FSHD-like phenotype**
Xingyu Xia, Wenhua Zhu, Chongbo Zhao

P4.02 **Comprehensive molecular study of patients with FSHD2**
Mikhail Skoblov, Darya Sherstyukova, Anna Orlova, Olga Shatokhina, Olga Shchagina, Anna Kuchina, Aysylu Murtazina

P4.03 **Evaluation of non-invasive biological sources for assessing methylation levels of FSHD-associated locus**
Domenica Megalizzi, Claudia Strafella, Giulia Trastulli, Emma Proietti Piorgo, Luca Colantoni, Raffaella Cascella, Francesca Torri, Mauro Monforte, Guido Primiano, Cristina Sancricca, Giorgio Tasca, Carlo Caltagirone, Giulia Ricci, Gabriele Siciliano, Enzo Ricci, Massimiliano Filosto, Emiliano Giardina

P4.04 **Interleukin 1 beta levels were significantly different in male FSHD patients**
Ceren Hangul, Simone Baldi, Filiz Ozcan, Sibel Berker Karauzum, Giulia Nannini, Elena Niccolai, Hilmi Uysal, Amedeo Amedei

P4.05 **Is *DUX4*-mediated hormone receptor dysregulation contributing to FSHD?**
Sabrina Pagnoni, Camila Simonetti, Nizar Y. Saad, Oliver King, Scott Harper, Alberto Rosa

POSTER PRESENTATIONS

Colorado Ballroom A–E

PEDIATRIC FSHD

P5.01 **Early-onset FSHD natural history and preclinical research pipeline at Melbourne Children's Campus**
Ian Woodcock, Chantal Coles, Peter Houweling, Katy de Valle

P5.02 **Lower body muscle MRI shows annual progression of fat fraction in Australian children with FSHD**
Ian Woodcock, Seth Friedman, Katy de Valle, Olivia DuCharme, Silvia Blemker, Jeff Statland

P5.03 **Methylation analyses of Australian children with FSHD**
Ian Woodcock, Katy de Valle, Chantal Coles, Peter Houweling, Peter Jones, Takako Jones

P5.04 **Outcomes to Validate Evaluations in Pediatric FSHD (MOVE Peds) – study outline**
Ian Woodcock, Natalie Katz, Seth Friedman, Katy Eichinger, Katy de Valle, Michaela Walker, Rebecca Clay, Doris Leung, Maya Hatch, Rabi Tawil, Jeff Statland

P5.05 **Preclinical models of childhood-onset FSHD**
Peter Houweling, Ian Woodcock, Chantal Coles, Katy de Valle, Richard Mills

CLINICAL STUDIES & TRIAL DESIGN

P6.01 **A retrospective cross-sectional clinical study identifies extracellular vesicle-associated circulating protein biomarkers for facioscapulohumeral muscular dystrophy**
Bilal Bayazit, Don Henderson, Rabi Tawil, Scott Harper, Nizar Y. Saad

P6.02 **Coproducing care quality standards in facioscapulohumeral muscular dystrophy (FSHD) in partnership with people with FSHD, carers, and healthcare professionals: A qualitative focus group study**
Enza Leone, Anand Pandyan, Alison Rogers, Richa Kulshrestha, Jonathan Hill, Fraser Philp

P6.03 **Deep phenotyping and comprehensive genetics characterization in atypical FSHD cases**
Giulia Ricci, Francesca Torri, Claudia Strafella, Liliana Vercelli, Giulio Gadaleta, Barbara Risi, Luca Colantoni, Emiliano Giardina, Tiziana Mongini, Massimiliano Filosto, Gabriele Siciliano

P6.04 **Increasing diversity in clinical trial participation: An exploration of clinical trial site engagement**
Luis Estevez, Ben Knisely, Trista Hardin, Erin Sandy, Haley Arellano, Rebecca Block, Jennifer Dunne, Kristi Clark

P6.05 **Safety and tolerability of losmapimod for the treatment of FSHD**
Mihaela Levitchi Benea, Vivekananda Ramana, John Jiang

P6.06 **The FSHD European Trial Network**
Ria de Haas, Sheila Hawkins, Richard Lemmers, Emiliano Giardina, Enrico Bugiardini, Elena Carraro, Julie Dumonceaux, Yann Péréon, Giorgio Tasca, Mauro Monforte, Nicol Voermans

P6.07 **The participants' perspective on clinical trials—a qualitative study**
Joost Kools, Lizan Stinissen, Wija Oortwijn, Nicol Voermans

P6.08 **The UK Facioscapulohumeral Muscular Dystrophy Patient Registry: A powerful tool to support clinical research and patient voice in the translational research pathway**
Helen Walker, Chiara Marini Bettolo, Robert Muni Lofra, Richard Orrell, Andrew Graham, Fiona Norwood, Mark Roberts, Tracey Willis, Emma Matthews, Mark Mencias, Kate Adcock

POSTER PRESENTATIONS

Colorado Ballroom A–E

LATE-BREAKING ABSTRACTS

P7.01 **Characteristics of the enrolled population in the Phase 3 REACH trial in facioscapulohumeral muscular dystrophy (FSHD): Preliminary results**
Nicol Voermans, Doris Leung, Marie-Helene Jouvin, John Jiang

P7.02 **Characterizing *SLC34A2* as a biomarker for FSHD**
Maria Traficante, Andrea O'Neill, Alexia Smith, Ujwala Pimparkar, Rabi Tawil, Jeffrey Statland, Robert Bloch

P7.03 **Small molecule augmentation of Notch signaling rescues models of DMD and FSHD**
Duc Dong, Shiv Kumar, Joseph Lancman, Sophie Hao

P7.04 **DUX4 protein partners in muscle cells are linked to DNA repair, transcription, and *DUX4* post-translational regulation**
Frédérique Coppée, Moriya Slavin, Clothilde Claus, Karimatou Bah, Keren Zohar, Tziona Eliyahu, Michal Linial, Nir Kalisman

P7.05 ***DUX4c* preservation: A key consideration in FSHD therapeutic strategies to safeguard muscle regeneration**
Clothilde Claus, Karimatou Bah, Moriya Slavin, Nir Kalisman, Alexandra Belayew, Frédérique Coppée

P7.06 **Reliability and validity of reachable workspace total score with wrist weights in facioscapulohumeral muscular dystrophy**
Lena Hubig, Adi Eldar-Lissai, Siu Hing Lo, John Jiang, Sarah Acaster, David Cella

P7.07 **Developing an updated standard of care and management for facioscapulohumeral muscular dystrophy**
June Kinoshita, Sarah Elmarkhous, Ronne Pater

P7.08 **A new home respiratory polygraphy for FSHD**
Patrick Valentin

P7.09 **An analysis of medical claims costs for individuals with FSHD in the United States**
June Kinoshita, Amanda Hill, Nicole Deypalan, Maryna Kolochavina, Man Hung, Eric Hon, Tanyatorn Ghanjanasak, Jamshid Arjomand

P7.10 **BetterLife FSHD: A new patient-driven health and research platform**
Amanda Hill, June Kinoshita

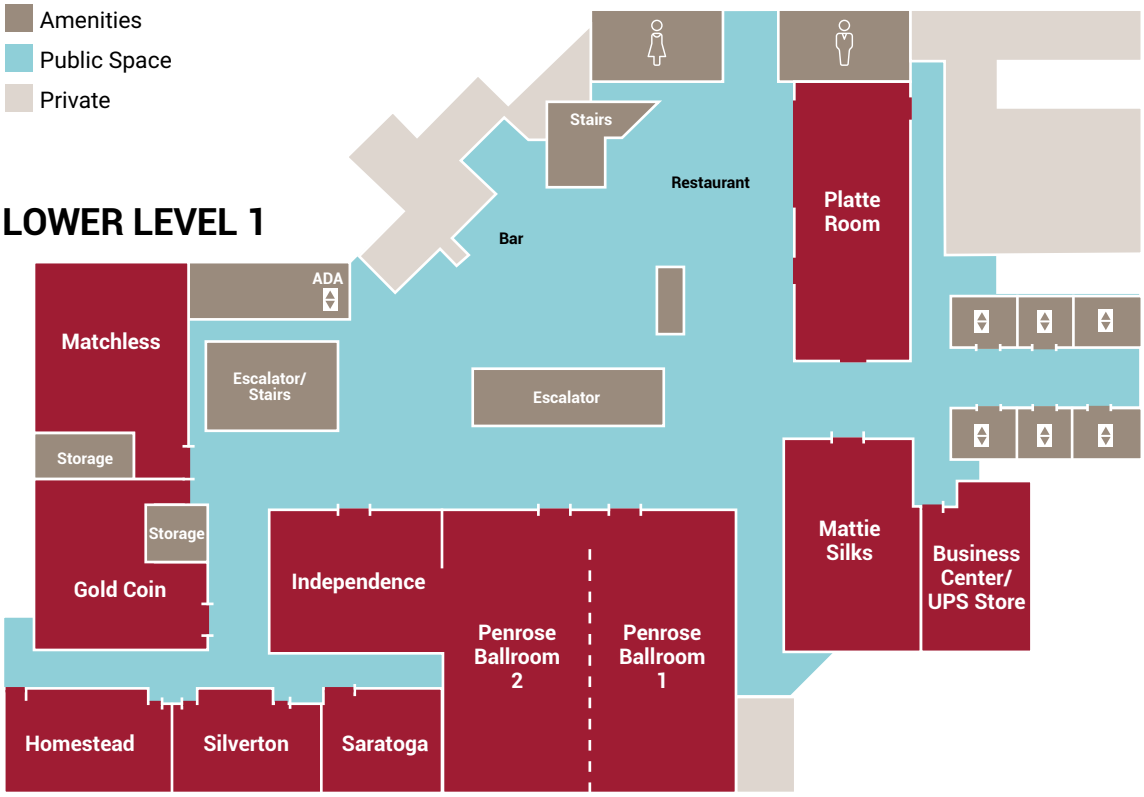
CONFERENCE AND EVENTS ROOMS

Floor Maps

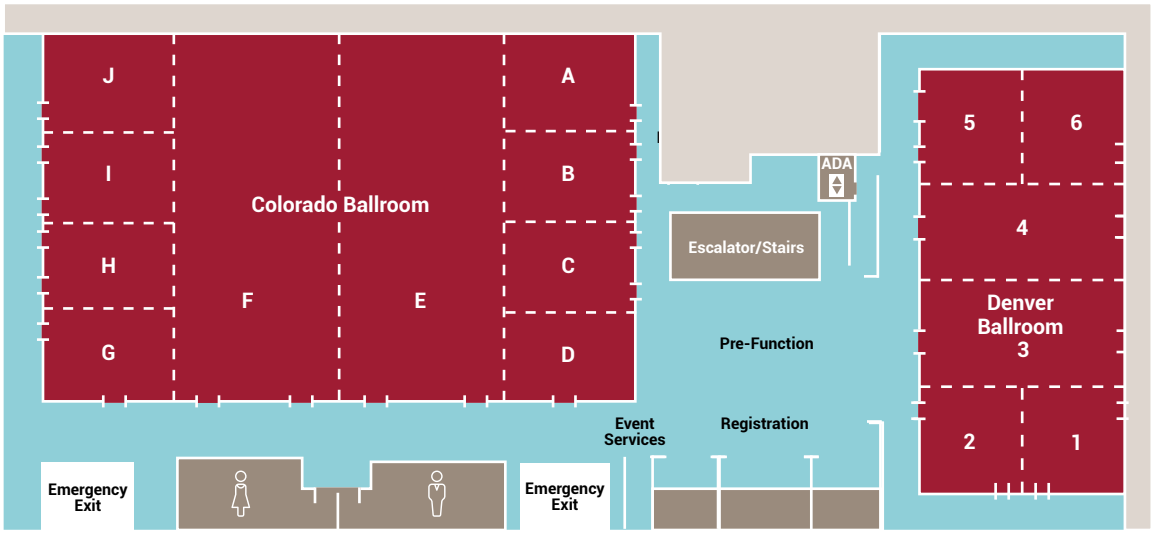
FLOOR MAPS KEY

- Meeting/Conference Rooms
- Amenities
- Public Space
- Private

LOWER LEVEL 1



LOWER LEVEL 2



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FSHD SOCIETY
LIGHTING THE WAY TO A CURE

Project Mercury

A new patient-driven global collaboration to speed the delivery of therapies for FSHD

AIMS

- Optimize clinical trial readiness
- Address barriers to patient access to treatments
- Facilitate productive collaboration in FSHD stakeholder ecosystem

PROJECT MERCURY

QUESTIONS? CONTACT:
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LEARN MORE AT
projectmercuryfshd.org