30th 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 15-16, 2023

Radisson Blu Hotel

Milan, Italy

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FSHD SOCIETY
LIGHTING THE WAY TO A CURE

BENVENUTI A TUTTI!

Welcome to the FSHD Society's 30th annual International Research Congress. This meeting, long established as the premier global platform for the discussion and dissemination of basic and clinical research on facioscapulohumeral muscular dystrophy (FSHD), is convening this year in the historic city of Milan. There will be a virtual livestream of the meeting for those who are not able to attend in person.

In the city where Leonardo da Vinci created some of history's greatest works in science, technology, and art, we will be sharing and discussing the latest advances in FSHD research. Ever since the genetic mechanism of FSHD was established by a sprawling international collaboration, our field has been moving at an ever-quickening pace toward treatments. There has never been a more urgent time to communicate and collaborate on behalf of patients and families.

This year, for the first time, we will devote a session to pediatric FSHD, a topic that has not received as much attention as FSHD in adults, even though most patients become symptomatic during childhood or adolescence. This is the age at which therapeutic interventions may yield the greatest benefit to patients, and so it is vitally important to deepen our understanding of this phase of FSHD. We will also hear more about research on the immune system's role, the genetics of FSHD in China and India, and many other topics ranging from basic discovery to clinical trials. We look forward to two days of stimulating talks and invigorating discussions as we re-dedicate ourselves to finding effective treatments for this devastating condition.

KEYNOTE SPEAKERS

Jaya Motta was born in Nepal in 2000. When he was only three years old, he was adopted by an Italian family and, since then, he has lived in Italy. Jaya is affected with FSHD and, for the last 12 years, he has used a wheelchair. He is currently a student studying biomedical engineering at the Politecnico di Milano. "I like engineering new ideas, creating new devices, but most of all I like helping people," he says. "This is why I've decided to take up my engineering studies. I want to combine my three most compelling desires, interests, and ambitions: 1) express my creativity, 2) take care of people, and 3) cure people." Jaya enjoys music, cinema, travel, and sports, in particular soccer and Formula One racing.

Baziel van Engelen, MD PhD, says "In my talk I will discuss some contributions of the Dutch FSHD expert center to the care and treatment of patients with FSHD. In addition to its successful translational approach, I will also highlight Miller Fisher's first rule: *The bedside can be your laboratory:* The capability approach in care, optimizing the trial toolkit in aerobic training and drug trials, and the potential of back-translational (bedside-to-bench) research."

Dr. van Engelen studied medicine at the Radboud University Medical Center and studied philosophy at the University of Amsterdam, which he finished cum laude in 1992. He did his neurology residency at the universities of Berlin and Nijmegen, followed by a research fellowship in the Mayo Clinic and Mayo Foundation in Rochester, Minnesota, USA. He became associate professor for neuromuscular diseases at the Radboud University Nijmegen Medical Center in 2000, full professor in 2003, and ENMC research director in 2010. He has published more than 250 articles in his research field. His focus is on clinical and translational research of neuromuscular disorders, especially myopathies.

Bénédicte Chazaud, PhD, is the director of research and team leader at the Institut NeuroMyoGène in Lyon, France. Her lab's research is dedicated to the role of environmental cells on adult muscle stem cell behavior in skeletal muscle regeneration. The Chazaud lab was a pioneer in bringing the concept of a stromal support that directly acts on muscle stem cells to regulate and promote an efficient muscle regeneration. The goal of their current studies is to decipher the molecular mechanisms underlying the cell interactions that allow proper myogenesis in normal muscle regeneration and during degenerating myopathies.

PROGRAM COMMITTEE

- Davide Gabellini, PhD (Co-chair, local host)
- Nicol Voermans, MD PhD (Co-chair)
- Jamshid Arjomand, PhD (Organizer)
- Alexandra Belayew, PhD
- Alberto Rosa, MD PhD
- Piraye Oflazer, MD
- Mauro Monforte, MD PhD
- Darko Bosnakovski, DVM PhD
- Katy Eichinger, PT DPT

Slide Presentations
DAY 1 – THURSDAY, JUNE 15, 2023

BLU CONFERENCE ROOM

9:00 a.m.
WELCOMING REMARKS

9:05 a.m.
KEYNOTE: Patient perspective on living with FSHD
Jaya Motta

9:35 a.m.
KEYNOTE: The Dutch FSHD expert center, translation and back-translation
Baziel van Engelen, MD PhD, Radboud University Medical Centre

10:30–11:00 a.m.
COFFEE BREAK AND NETWORKING
Foyer Blu

11:00 a.m.–noon
SESSION 1: Discovery Research & Genetics
Session Chairs: Davide Gabellini, PhD, & Darko Bosnakovski, DVM PhD

11:00 a.m.
S1.01 FSHD-like muscle pathology in a mouse *Dux* inducible transgenic mouse model
Darko Bosnakovski, University of Minnesota

11:20 a.m.
S1.02 Activation of the germline transcription factor *DUX4* is essential for herpesvirus replication
Florian Full, University Medical Center, Freiburg

11:40 a.m.
S1.03 SIX transcription factors promote differentiation-dependent activation of *DUX4* expression in FSHD
Amelia Fox, Saint Louis University

12:00 p.m.
LUNCH BREAK
Leonardo Restaurant

12:30–2:00 p.m.
Poster session and networking. Odd-numbered poster presentations.
Meeting Rooms 9 & 10, and Foyer Blu

2:00–3:00 p.m.
SESSION 1 PART 2: Discovery Research & Genetics

2:00 p.m.
S1.04 Long-read sequencing reveals novel transcripts induced by misexpression of *DUX4* in FSHD muscle
Dongxu Zheng, Leiden University

2:20 p.m.
S1.05 Candidate circulating biomarkers for FSHD
Joel Chamberlain, University of Washington

2:40 p.m.
S1.06 Molecular diagnosis of FSHD1 in India suggests a lower clinical susceptibility compared to patients with a European background
Richard Lemmers, Leiden University

3:00 p.m.
COFFEE BREAK AND NETWORKING
Foyer Blu

3:30–5:30 p.m.
SESSION 2: Outcome Assessments
Session Chairs: Kate Eichinger, PT DPT, & Mauro Monforte, MD PhD

3:30 p.m.
S2.01 Disease progression in FSHD1 patients with different D4Z4 methylation levels: a Chinese follow-up study
Zhiqiang Wang, First Affiliate Hospital of Fujian Medical University

3:50 p.m.
S2.02 Identification of pharmacodynamic and monitoring biomarkers for facioscapulohumeral muscular dystrophy
Yi-Wen Chen, Children's National Research Institute

4:10 p.m.
S2.03 Muscle imaging in natural history of FSHD: Quantitative MRI and ultrasound results compared head-to-head
Sanne Vincenten, Radboud University Medical Center

4:30 p.m.
S2.04 Evaluation of disease progression in facioscapulohumeral muscular dystrophy using multiparametric MRI
Mauro Monforte, MD PhD, Fondazione Policlinico Universitario A. Gemelli IRCCS

4:50 p.m.
S2.05 Motor outcomes to validate evaluations in facioscapulohumeral muscular dystrophy (MOVE FSHD): Preliminary baseline characteristics
Michaela Walker, University Kansas

5:10 p.m.
S2.06 Facioscapulohumeral muscular dystrophy (FSHD) surgeries, cardiovascular testing, mobility aids and healthcare utilization after diagnosis from a real-world data analysis
Elizabeth Ackermann, Avidity Biosciences

5:30 p.m.
Adjourn

6:30 p.m.
Reception
Blu Conference Room Foyer

7:30 p.m.
Banquet
Blu Conference Room

Slide Presentations
DAY 2 – FRIDAY, JUNE 16, 2023

BLU CONFERENCE ROOM

9:00 a.m.
WELCOMING REMARKS

9:05 a.m.
KEYNOTE: Macrophages during skeletal muscle regeneration: Friend or foe?
Bénédicte Chazaud, PhD, Institut NeuroMyoGène

10:05–10:30 a.m.
COFFEE BREAK AND NETWORKING
Foyer Blu

10:30 a.m.–12:30 p.m.
SESSION 3: Disease Mechanisms & Interventional Strategies
Session Chairs: Alexandra Belayew, PhD, & Alberto Rosa, MD PhD

10:30 a.m.
S3.01 Immunopathogenesis of facioscapulohumeral muscular dystrophy (FSHD)
Beatrice Biferali, San Raffaele Institute

10:50 a.m.
S3.02 An innate immune cell/FSHD muscle xenograft model to investigate the role of complement pathway activation in FSHD muscle pathology
Katelyn Daman, UMass Chan Medical School

11:10 a.m.
S3.03 Regulation of muscle regeneration through FSHD disease progression
Elise Engquist, King's College London

11:30 a.m.
S3.04 *DUX4* and *DUX4c* directly interact with C1qBP in FSHD regenerating myofibers
Clothilde Claus, University of Mons

11:50 a.m.
S3.05 The interactome of *DUX4* reveals an inherent feedback mechanism by RFPL4
Moriya Slavin, Hebrew University

12:10 p.m.
S3.06 Developing Cas13-ADAR-mediated *DUX4* mRNA editing as a prospective therapy for FSHD
Scott Harper, Nationwide Children's Hospital

12:30 p.m.
LUNCH BREAK
Leonardo Restaurant

1:00–2:25 p.m.
Poster session and networking. Even-numbered posters presenting.
Meeting Rooms 9 & 10, and Foyer Blu

2:25–2:30 p.m.
Vote for Best Poster

2:30–3:30 p.m.
SESSION 4: Clinical Studies & Trial Designs
Session Chairs: Nicol Voermans, MD PhD, & Piraye Oflazer, MD

2:30 p.m.
S4.01 Results from 96-week open-label extension of a Phase 2 trial of losmapimod in subjects with FSHD: ReDUX4 – overview of REACH
Marie-Helene Jouvin, Fulcrum Therapeutics, & Leo Wang, MD, University of Washington

2:45 p.m.
S4.02 Phase 1/2 trial evaluating AOC 1020 in adults with FSHD: FORTITUDE trial
Amy Halseth, Avidity Biosciences

3:00 p.m.
S4.03 MANOEUVRE study design: A study of GYM329 (RO7204239) in patients with facioscapulohumeral muscular dystrophy (FSHD)
Giorgio Tasca, JWMDRC, Newcastle University

3:15 p.m.
S4.04 PERSPECTYV FSHD: PERsonalized Medicine and SPECialized TherapY for better LiVing with FSHD
Dalila Laoudj-Chenivesse, INSERM

3:30–4:00 p.m.
COFFEE BREAK AND NETWORKING
Foyer Blu

4:00–5:10 p.m.
SPECIAL SESSION: Pediatric FSHD
Chair: Nicol Voermans, MD PhD

4:00–4:20 p.m.
S5.01 Dutch pediatric study in childhood FSHD
Corrie Erasmus, Radboud University Medical Centre

4:20–4:40 p.m.
S5.02 Measuring function in childhood FSHD: Does the FSHD-COM Peds measure up?
Katy de Valle, Royal Children's Hospital, Melbourne

4:40–5:10 p.m.
Panel Discussion on Pediatric FSHD
Chaired by Nicol Voermans, MD PhD
Panelists: Corrie Erasmus, Jeffrey Statland, Katy de Valle, Ian Woodcock

5:10–5:25 p.m.
Best Poster Award & Young Investigator Award
Davide Gabellini, PhD, and Nicol Voermans, MD PhD

5:25 p.m.
2024 IRC announcement & final remarks

5:30 p.m.
Adjourn

Poster Presentations

*Odd-numbered posters will be presented on **June 15**. Even-numbered posters will be presented on **June 16.***

DISCOVERY RESEARCH & GENETICS

P1.01 **Dominant pathogenic cis D4Z4 repeat duplications in FSHD**
Richard Lemmers, Russell Butterfield, Patrick J. van der Vliet, Robert B. Weiss, Silvère van der Maarel, Jan De Bleecker, Ludo van der Pol, Corrie Erasmus, Baziel van Engelen, Nicol Voermans, Marc D'Hooghe, Kristof Verhoeven, **Jeff Statland,** Enrico Bugiardini, Nienke van der Stoep, Teresinha Evangelista, Chiara Marini-Bettolo, Peter van den Bergh, Rabi Tawil, John Vissing, Judit Balog

P1.02 **Whole Exome Sequencing of 126 patients provides evidence for novel candidate genes in FSHD**
Claudia Strafella, Domenica Megalizzi, Valerio Caputo, Giulia Trastulli, Luca Colantoni, Sara Bortolani, Eleonora Torchia, Mauro Monforte, Carlo Caltagirone, Enzo Ricci, Giorgio Tasca, Emiliano Giardina, Raffaella Cascella

P1.03 **Do the novel mutations in FSHD point to polygenic disease? Co-segregated *MYH2* and *GP1BA* gene mutations in a FSHD family with hereditary thrombocytopenia**
Ceren Hangul, Haldun Dogan, Sibel Berker Karauzum, Hilmi Uysal, Serdar Ceylaner

P1.04 **Optical genome mapping for the molecular diagnosis of facioscapulohumeral muscular dystrophy: Advancements and challenges**
Stephanie Efthymiou, VY Vishnu, Richard J. F. L. Lemmers, Lindsay Wilson, Patrick J. van der Vliet, Natalia Dominik, Perrone Benedetta, Stefano Facchini, Andrea Cortese, Silvère van der Maarel, Michael Hanna, Enrico Bugiardini

P1.05 **D4Z4 methylation analysis combined with machine learning pipelines: a novel tool for identifying FSHD subjects**
Raffaella Cascella, Valerio Caputo, Claudia Strafella, Giulia Trastulli, Domenica Megalizzi, Carlo Fabrizio, Andrea Termine, Luca Colantoni, Juliette Gimenez, Mauro Monforte, Carlo Caltagirone, Enzo Ricci, Giorgio Tasca, Emiliano Giardina

P1.06 **MethylSeq-based assay to assess the epigenetic setting of D4Z4 repetitive elements in facioscapulohumeral muscular dystrophy**
Valentina Salsi, **Sara Pini,** Matteo Chiara, Lucia Ruggiero, Filippo Santorelli, Stefano Previtali, Diego Lopergolo, Maria Grazia D'Angelo, Carmelo Rodolico, Silvia Bonanno, Lorenzo Maggi, Rossella Tupler

P1.07 **DUX4 protein interactors are involved in the DNA damage response**
Karimatou Bah, Moriya Slavin, Clothilde Claus, Anne-Emilie Declèves, Nir Kalisman, Frédérique Coppée

P1.08 **SMCHD1 regulates biological pathways relevant for Bosma syndrome and facioscapulohumeral dystrophy phenotype**
Frédérique Magdinier, Camille Laberthonniere

P1.09 **A study of *DUX4* expression pattern with FSHD patient-derived iPSC model**
Mitsuru Sasaki-Honda, Álvaro Rada-Iglesias, Hidetoshi Sakurai

P1.10 **Fibro-adipogenic progenitors and FSHD myopathy**
Carlo Serra, Kathryn Wagner, Andrew Wilson, Thomas Lloyd

P1.11 ***DUX4*, nucleolar stress, apoptosis, and FSHD myopathy**
Carlo Serra, Kathryn Wagner, Andrew Wilson, Thomas Lloyd

P1.12 **Deciphering the role of SMCHD1 in disease and development**
Rachel Eiges, Uria Aviel, Silvina Epsztein-Litman, Yotam Drier

P1.13 **Generation of mouse artificial chromosome carrying FSHD1-derived chromosome 4q35 for a novel FSHD1 mouse model**
Yosuke Hiramuki, Ichizo Nishino, Hiroyuki Kugoh, Yasuhiro Kazuki

P1.14 **Development of a new *DUX4*-responsive reporter mouse**
Lindsay Wallace, Jessica Camp, Noah Taylor, Scott Q. Harper

P1.15 **FRG1-mouse as an effective model of muscle wasting to test novel therapeutic options for FSHD**
Sebastian Fantini, Grazia Bisceglia, Beatrice Bertarini, Rui Li, Marcello Manfredi, Mathieu Michel, Marco Spinazzi, Lihua Zhu, Pascal Reynier, Giuseppe D'Antona, Rossella Tupler

P1.16 **FSHD 3D-modeling through different bioprinting approaches**
Stefano Testa, Lucas Duvert, Patricia Alloncle, Adrien Casanova, Frédérique Magdinier

P1.17 **WDR5 is required for *DUX4* expression and its pathological effects in FSHD muscular dystrophy**
Emanuele Mocciaro, Roberto Giambruno, Stefano Micheloni, Filippo Cernilogar, Annapaola Andolfo, Cristina Consonni, Maria Pannese, Giulia Ferri, Valeria Runfola, Gunnar Shotta, Davide Gabellini

P1.18 **Discordant monozygotic twins with reduced-length D4Z4 allele and FSHD-like phenotype**
Giulio Gadaleta

OUTCOMES ASSESSMENTS

P2.01 **Assessment of the burden of outpatient clinic and MRI-guided needle muscle biopsies as reported by patients with facioscapulohumeral muscular dystrophy**
Joost Kools, Willem Aerts, Erik Niks, Karlien Mul, Lisa Pagan, Jake Maurits, Renée Thewissen, Baziel van Engelen, Nicol Voermans

P2.02 **Development and validation of the facioscapulohumeral muscular dystrophy-health index (FSHD-HI), a disease-specific patient-reported outcome measure to facilitate clinical trials**
Anika Varma, Jennifer Weinstein, Jamison Seabury, Spencer Rosero, Charlotte Engebrecht, Ellen Wagner, Christine Zizzi, Elizabeth Luebbe, Nuran Dilek, Michael McDermott, John Kissel, Valeria Sansone, Chad Heatwole

P2.03 **A systematic literature review to assess the level of evidence in facioscapulohumeral muscular dystrophy**
Lianne Barnieh, Rachel Beckerman, Helena Emich, Katy Eichinger, Adi Eldar-Lissai

P2.04 **Developing a therapist-led FSHD clinic at the Atkinson-Morley Neuromuscular Centre**
Sherryl Chatfield, Pamela Appleton, Niranjanan Nirmalananthan, Emma Matthews

P2.05 **Muscle diffusion tensor imaging in facioscapulohumeral muscular dystrophy**
Leonardo Barzaghi, Matteo Paoletti, Mauro Monforte, Sara Bortolani, Chiara Bonizzoni, Thorsten Feiweier, Niels Bergsland, Francesco Santini, Xeni Deligianni, Giorgio Tasca, Silvia Figini, Enzo Ricci, Anna Pichiecchio

P2.06 **Exploring the use of facial muscle ultrasound in facioscapulohumeral muscular dystrophy**
Sjan Teeselink, David Lamers, Sanne Vincenten, Nens van Alfen, Baziel van Engelen, Nicol Voermans, Karlien Mul

P2.07 **Facioscapulohumeral muscular dystrophy (FSHD) age-related differences in management among patients over and under 40 years**
Kathryn Munoz, Chamindra Laverty, Richard Brook, Nathan L. Kleinman, Chao-Yin Chen, Teresa Brandt, Mark C. Stahl, **Amy Halseth**

P2.08 **FSHD European Trial Network**
Nicol Voermans, Valeria Sansone, Giorgio Tasca, Federica Montagnese, Karlien Mul, Richard J. F. L. Lemmers, Emiliano Giardina, Pascal Laforet, María Vriens-Munoz Bravo, Alexandre Mejat, Julie Dumonceaux, Teresinha Evangelista

P2.09 **Respiratory tests and models in FSHD**
Patrick Valentin

P2.10 **Standard of care and management of facioscapulohumeral muscular dystrophy**
Ronne Pater, Sarah el Markhous, June Kinoshita

DISEASE MECHANISMS & INTERVENTIONAL STRATEGIES

P3.01 **Characterization of the D4Z4 subtelomeric region of a human derived isogenic iPSC line and identification of a CRISPR/Cas9 strategy for *DUX4* inactivation in facioscapulohumeral muscular dystrophy type 1 (FSHD1)**
Edoardo Malfatti, Chéryane Lama, Nicolas de Graaf, Reem Bou Akar, Prisca Danaus, Laurence Suel-Petat, Juliette Nectoux, François-Jérôme Authier, Frédéric Relaix, Isabelle Richard

P3.02 **EPI-321: A promising gene therapy for facioscapulohumeral muscular dystrophy (FSHD) targeting D4Z4 epigenome**
Abhinav Adhikari, Alexandra Collin de l'Hortet

P3.03 **Identification of the first direct endogenous inhibitor of *DUX4* in FSHD muscular dystrophy**
Valeria Runfola, Paola Ghezzi, Maria Pannese, Roberto Giambruno, Claudia Caronni, Annapaola Andolfo, Davide Gabellini

P3.04 ***ANT1* overexpression models: Phenotype similarities with FSHD**
Sandrine Arbogast, Heinrich Kotzur, Corinna Frank, Nathalie Compagnone, Thibault Sutra, Fabien Pillard, Sylvia Pietri, Nisrine Hmada, Daouda Moustapha Abba Moussa, Jacques Mercier, Jean-Paul Cristol, Marie-Christine Dabauvalle, Dalila Laoudj-Chenivesse

P3.05 **Apabetalone, a clinical-stage, selective BET inhibitor, opposes DUX4 expression in primary human FSHD muscle cells**
Ewelina Kulikowski, Christopher Sarsons, Dean Gilham, Laura Tsujikawa, Li Fu, Sylwia Wasiak, Brooke Rakai, Stephanie Stotz, Agostina Carestia, Michael Sweeney, Jan Johansson, Norman Wong

P3.06 **Sustained efficacy of CRISPR-Cas13b gene therapy for FSHD is challenged by immune response to AAV.Cas13b vectors**
Scott Q. Harper, Afrooz Rashnonejad, Gerald Coulis, Noah Taylor, Gholamhossein Amini-Chermahini, Armando Villalta, Oliver King

P3.07 **A modular system to convert therapeutic miRNAs from ubiquitous RNA pol III-based promoters to RNA pol II-driven muscle-specific promoters while maintaining fidelity of processing and efficacy**
Noah Taylor, Matthew Guggenbiller, Lindsay Wallace, Scott Q. Harper

P3.08 **Safety and efficacy of a possible gene therapy approach for FSHD muscular dystrophy**
Maria Pannese, Beatrice Biferali, Ellen Wiedtke, Valeria Runfola, Paola Ghezzi, Dirk Grimm, Davide Gabellini

P3.09 ***DUX4*-mediated hypoxia signaling and impairment of oxygen metabolism in facioscapulohumeral muscular dystrophy**
Justin Cohen, Vincent Ho, Alec Desimone, Keryn Woodman, Monkol Lek

P3.10 **Selection of peptides for a muscle-targeted delivery of ASO 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, Alexandra Belayew, Frédérique Coppée, Anne-Emilie Declèves, Alexandra Tassin

P3.11 **Estrogen interferes with *DUX4* nuclear import**
Giada Mele, Fabiola Moretti, Giancarlo Deidda, Patrizia Calandra, Emanuela Teveroni, Silvia Maiullari, Marco Crescenzi, Maria Luisa Casella

P3.12 **WITHDRAWN**

P3.13 **Unravelling the contribution of non-myogenic mesenchymal cells in the pathogenesis of facioscapulohumeral muscular dystrophy**
Lorena Di Pietro, Andrea Papait, Elvira Ragozzino, Antonietta Silini, Pietro Romele, Wanda Lattanzi, Enzo Ricci, Ornella Parolini

P3.14 **Reversing the altered behavior of non-myogenic mesenchymal cells from FSHD patients: Potential of human amniotic cell conditioned medium**
Flavia Giacalone, Lorena Di Pietro, Andrea Papait, Elvira Ragozzino, Antonietta Silini, Pietro Romele, Wanda Lattanzi, Enzo Ricci, Ornella Parolini

P3.15 **A retrospective cohort study identifies fibrosis as candidate biomarker for muscle degeneration in facioscapulohumeral muscular dystrophy patients**
Elvira Ragozzino, Sara Bortolani, Lorena Di Pietro, Andrea Papait, Ornella Parolini, Mauro Monforte, Giorgio Tasca, Enzo Ricci

P3.16 **Retrospective analysis of muscle biopsy findings in a cohort of patients with facioscapulohumeral dystrophy type 1**
Lucia Ruggiero, Matthew Guggenbiller, Lindsay Wallace, Scott Q. Harper

P3.17 **Symptom onset and cellular pathology in facioscapulohumeral muscular dystrophy is accelerated by cigarette smoking**
Christopher Banerji, Matthew Guggenbiller, Lindsay Wallace, Scott Q. Harper

P3.18 **Single cell proportion analysis identifies unique transcriptional responses to plasma membrane injury in FSHD**
Adam Bittel, Matthew Guggenbiller, Lindsay Wallace, Scott Q. Harper

P3.19 **Facioscapulohumeral disease as a myodevelopmental disease: Applying Ockham's razor to its various features**
Nicol Voermans, George Padberg, Baziel van Engelen

P3.20 **Developmental repression/derepression of *DUX4* 4qA during FSHD embryonic and adult myogenesis**
Dongsheng Guo, Katelyn Daman, Jing Yan, Lawrence Hayward, Oliver King, **Charles Emerson**

P3.21 **An in silico FSHD muscle fibre for modelling *DUX4* dynamics and predicting the impacts of therapy**
Matthew Cowley, Johanna Pruller, Massimo Ganassi, Peter Zammit, Christopher Banerji

P3.22 **AOC 1020: An antibody oligonucleotide conjugate (AOC) in development for the treatment of FSHD**
Barbora Malecova, David Sala, Garineh Mary Melikian, Nathan Delos Santos, Gulin Erdogan, Rachel Johns, Maryam Jordan, Marc Hartmann, Danny Arias, Arvind Battacharya, Ramana Doppalapudi, Hanhua Huang, Michael Flanagan, Arthur Levin

CLINICAL STUDIES & TRIAL DESIGNS

P4.01 **Safety and tolerability of losmapimod for the treatment of FSHD**
Marie-Helene Jouvin, Vivekananda Ramana, John Jiang

P4.02 **Effect of creatine monohydrate on functional muscle strength and muscle mass in children with FSHD: A multi-centre, randomised, double-blind placebo-controlled crossover trial**
Ian Woodcock, Katy de Valle, Anita Cairns, Nisha Varma, Martin Delatycki, Eppie Yiu, Zoe Davidson, Michael Kean, Aneke Grobler

P4.03 **Building an integrated machine learning-based platform to study FSHD: From deep phenotyping to predictive biomarkers**
Giulia Ricci, Stefano Cotti Piccinelli, Francesca Torri, Giulio Gadaleta, Roberto Gatta, Emanuele Frontoni, Federica Decorato, Stefano Regondi, Alessandro Tonacci, Francesco Sansone, Raffaele Conte, Alessandro Padovani, Tiziana Mongini, Gabriele Siciliano, Massimiliano Filosto

P4.04 **Prescription of pain medication for people with facioscapulohumeral muscular dystrophy in the Muscular Dystrophy Surveillance, Tracking, and Research Network**
Kristin Conway, Jonathan Suhl, Katherine Mathews, Sonja Rasmussen, James Howard Jr., Jennifer Andrews, Shiny Thomas, Nicholas Johnson, Joyce Alese, Paul Romitti

P4.05 **Diaphragmatic ultrasound: A promising technique for respiratory assessment of patients with facioscapulohumeral muscular dystrophy (FSHD)**
Eleonora Torchia, Sara Bortolani, Riccardo Inchingolo, Andrea Smargiassi, Mauro Monforte, Giorgio Tasca, Matteo Bonini, Luca Richeldi, Enzo Ricci

P4.06 **Quality of life and support needs in children and adolescents with facioscapulohumeral dystrophy: A qualitative study**
Jildou Dijkstra, Nicol Voermans, Corrie Erasmus, Baziel van Engelen, Edith Cup, Sandra Altena, Eline Boon, Nathaniël Rasing, Anke Lanser, Anouska Ramaker

P4.07 **Clinically Relevant Outcome Measures in FSHD (CROMFiSH): Results of a twelve-month longitudinal natural history study**
Anika Varma, Katy Eichinger, Michael Todinca, Susanne Heininger, Nuran Dilek, Bill Martens, John Kissel, Rabi Tawil, **Jeff Statland,** Michael McDermott, Chad Heatwole

P4.08 **FSHD European Patient Survey: Assessing patient preferences in clinical trial participation**
Magan McNiff, Sheila Hawkins, Bine Haase, Joanne Bullivant, Tammy McIver, Olga Mitelman, Nicholas Emery, Giorgio Tasca, Nicol Voermans, Jordi Diaz-Manera

P4.09 **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, Richard Orrell, Andrew Graham, Fiona Norwood, Mark Roberts, Tracey Willis, Emma Matthews, Mark Mencias, Kate Adcock, Robert Muni-Lofra

P4.10 **FSHD UK: Creating a strong multi-stakeholder group to strategically drive clinical trial readiness and co-ordination of FSHD activities in the UK. On behalf of FSHD UK**
Rajeshri Badiani

LATE-BREAKING ABSTRACTS

P5.01 **Best practice guidelines on genetic diagnostics of facioscapulohumeral muscular dystrophy (FSHD): Update of the 2021 guidelines**
Emiliano Giardina, Pilar Camaño, Nienke van der Stoep, Valerie Race, Sara Burton-Jones, Victoria Williams, Silvère M. van der Maarel, Nicol Voermans, Richard J. F. L. Lemmers

P5.02 **True cost of FSHD: Health economic study of facioscapulohumeral muscular dystrophy in the United States**
June Kinoshita**, Amanda Hill**, Maryna Kolochavina, Man Hung, Nicole Deypalan, Eric Hon, Jamshid Arjomand

P5.03 **TestFSHD: A fully sponsored direct-to-patient clinically approved genetic testing pilot program for US patients**
June Kinoshita, Leigh Reynolds, **Jamshid Arjomand**

P5.04 **Physical interaction between *DUX4* and hormone nuclear receptors**
Sabrina Pagnoni, Julieta Quintero, Nizar Y. Saad, Scott Q. Harper, Alberto L. Rosa

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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:
Ken Kahtava — *ken.kahtava@fshdsociety.org*
Mark Stone, Chair, Global Task Force — *mark.stone@fshdsociety.org*

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