PRELIMINARY PROGRAM

June 12-13, 2025
Leonardo Royal Hotel
Amsterdam, the Netherlands

DAY 1: THURSDAY, JUNE 12, 2025

Amstel 1 & 2

8:00–9:00 a.m.
REGISTRATION
Breakfast on your own

9:00 a.m.
WELCOME
Lucienne Ronco, organizing chair, & Stephen Tapscott, co-chair

9:05 a.m.
KEYNOTE: *Marion and the Metamorphosis* (film excerpt)
Marion Sellenet, artist living with FSHD

9:20 a.m.
KEYNOTE: Antibodies and complement, partners in crime but also opportunities to intervene
Leendert Trouw, Leiden University Medical Center

In many autoimmune and inflammatory conditions, the presence of antibodies triggers activation of the complement system, a protein-based part of the innate immune defense. Understanding the nature of the antibody response and the targets to which the antibodies bind allows insight into the disease-driving processes. Ascertaining the processes by which the antibodies contribute to tissue pathology, activation of complement, triggering of cellular Fc-receptors, or blocking of physiological functions provides insight into the therapeutic options available to address the immunopathology. In this opening lecture, parallels and differences will be discussed among autoimmune diseases like rheumatoid arthritis, systemic lupus erythematosus, and facioscapulohumeral muscular dystrophy. In addition, new and exciting options will be discussed on therapeutic intervention focusing on antibodies and complement.

SLIDE PRESENTATIONS

SESSION 1: Population Genetics & Modifiers

Session Chairs: Christiane Moreno & Vishnu VY

10:00 a.m.
S1.01 FSHD in sub-Saharan Africans
Richard Lemmers, Leiden University Medical Center

10:20 a.m.

S1.02 Analysis of genetic diversity and phylogenetic relationships of D4Z4 repeats: Implication for health and disease
Sara Pini, University of Modena, & Reggio Emilia, Modena

10:40 a.m.
S1.03 Correlation of methylation, severity, and parent-of-origin effects in large, multigenerational kindred with FSHD
Russell Butterfield, University of Utah

11:00 a.m.
S1.04 Exploring the boundaries of the diagnostic spectrum of FSHD: Complex genetic findings and their implication for the molecular genetic model of the disease
Hannes Erdmann, Medical Genetics Center Munich and Friedrich-Baur-Institut at the Department of Neurology, Ludwig Maximilian University Hospital, Munich, Germany

11:20–11:40 a.m.
COFFEE BREAK & NETWORKING
Foyer

SESSION 2: Measures of Disease Activity & Progression

Session Chairs: Seth Friedman & Giorgio Tasca

11:40 a.m.
S2.01 FSHD1 in Italy: A 20-year follow-up study of the Italian Clinical Group for FSHD (AIM-FSHD)
Giulio Gadaleta, University of Turin

12:00 p.m.
S2.02 Integrating MRI, RNAseq, and pathology to predict fat fraction change over 1 year in FSHD
Seth Friedman, Seattle Children's Hospital

12:20 p.m.
S2.03 Multi-scale machine learning model predicts muscle and functional disease progression in FSHD
Silvia Blemker, University of Virginia and Springbok Analytics

12:40–1:40 p.m.
LUNCH BREAK
LEO's International Flavors

SESSION 2: Measures of Disease Activity & Progression (continued)

Session Chairs: Seth Friedman & Giorgio Tasca

1:40 p.m.
S2.04 Identification of potential plasma extracellular vesicle-associated protein biomarkers for facioscapulohumeral muscular dystrophy (FSHD)
Nizar Saad, Nationwide Children's Hospital and The Ohio State University College of Medicine

2:00 p.m.
S2.05 Myostatin is a biomarker of disease severity in facioscapulohumeral muscular dystrophy
Julie Dumonceaux, University College London

2:20 p.m.
S2.06 Extracellular vesicle RNA profiling reveals candidate biomarkers of disease activity and progression for FSHD patients
Elvira Ragozzino, Università Cattolica del Sacro Cuore

2:40–3:00 p.m.
COFFEE BREAK & NETWORKING
Foyer

SESSION 3: Novel Clinical Outcome Measures

Session Chairs: Enrico Bugiardini & Jeffrey Statland

3:00 p.m.
S3.01 Deep learning-based facial movement analysis for automated detection and severity classification of facioscapulohumeral muscular dystrophy (FSHD)
Sabrina Sacconi, Nice University Hospital and Institute for Research on Cancer and Aging of Nice

3:20 p.m.
S3.02 Reassessing training thresholds in FSHD: The diagnostic value of compensatory movement patterns
Nicole Voet, Radboud University Medical Center and Klimmendaal, Rehabilitation Center, Arnhem, Netherlands

3:40 p.m.
S3.03 Current abilities score (CAS) is a valid and a reliable patient-reported functional rating scale in FSHD: Data from MOVE natural history study
Channa Hewamadduma, University of Sheffield

4:00 p.m.
S3.04 Gait-based biomarkers in facioscapulohumeral dystrophy (FSHD): Establishing the validity of inertial measurement units (IMU) to augment in clinic assessments in FSHD
Jon Street, University of Sheffield

4:20–5:00 p.m.
POSTER MINI-TALKS

5:00 p.m.
CLOSING REMARKS & ONE-HOUR BREAK

EVENING SESSION

6:00–6:30 p.m.
POSTER MINI-TALKS
Amstel 1 and 2

6:30–8:00 p.m.
"WALKING DINNER" & POSTER SESSION
Amstel 3

8:00–9:00 p.m.
FILM SCREENING: ***Marion and the Metamorphosis***
Amstel 1 & 2

DAY 2: FRIDAY, JUNE 13, 2025

Amstel 1 & 2

8:00–9:00 a.m.
REGISTRATION
Breakfast on your own

9:00 a.m.
WELCOME
Lucienne Ronco, organizing chair, & Nicol Voermans co-chair

9:05 a.m.
KEYNOTE: A patient's perspective: Challenges, involvement, and future directions
Mart Brandjes

9:20–10:00 a.m.
KEYNOTE: Reflections on the development of therapies for spinal muscular atrophy
Karen Chen, SMA Foundation

Spinal muscular atrophy (SMA) is a rare neuromuscular genetic disease in which the degeneration of motor neurons and intrinsic muscle defects result in muscle atrophy and weakness. In the last eight years, the US Food and Drug Administration has approved three drugs: An antisense oligonucleotide, a gene therapy, and a small molecule that all address the genetic cause of SMA, stabilizing patients and preventing further degeneration. However, there is still a significant unmet need in patients, especially older patients. To address this, approaches that target improving muscle function that could be used in combination with the approved treatments are being developed. The presentation will provide an overview of the strategies and tools that enabled the development of SMA drugs from the perspective of the SMA Foundation, a nonprofit organization devoted to therapeutic development, as well as learning that could be applied to other diseases such as FSHD, and future directions.

SLIDE PRESENTATIONS

SESSION 4: Mechanisms of Disease & Interventional Strategies

Co-chairs: Marnie Blewitt & Kyoko Yokomori

10:00 a.m.
S4.01 SUMOylation regulates SMCHD1 activity and DUX4 expression in FSHD muscle
Judit Balog, Leiden University Medical Center

10:20 a.m.
S4.02 Mechanism of SMCHD1 and engineering for FSHD treatment
Shifeng Xue, National University of Singapore

10:40 a.m.
S4.03 Understanding and treating inflammation in FSHD muscular dystrophy
Beatrice Biferali, San Raffaele Scientific Institute

11:00–11:20 a.m.
COFFEE BREAK & NETWORKING
Foyer

SESSION 4: Mechanisms of Disease & Interventional Strategies

(continued)

Co-chairs: Marnie Blewitt & Kyoko Yokomori

11:20 a.m.
S4.04 Single-cell RNA sequencing identifies ferroptotic stress in FSHD myoblasts
Adam Bittel, Children's National Hospital

11:40 a.m.
S4.05 Decoding the cellular and epigenetic landscape of affected and spared muscles in FSHD patients through single nucleus multi-omic analysis
Lorena Di Pietro, Dipartimento di Scienze della Vita e Sanità Pubblica, Università Cattolica del Sacro Cuore, Rome, Italy, and Fondazione Policlinico Universitario A. Gemelli IRCSS

12:00 p.m.
S4.06 DYNE-302 leads to functional improvement and resolves muscle transcriptomic changes in mouse models of FSHD
Stefano Zanotti, Dyne Therapeutics

12:20 p.m.
S4.07 SRK-015 improves muscle mass, strength, and endurance in the FLExDUX4.Cre mouse model of FSHD
Adam Fogel, Scholar Rock

12:40 p.m.
S4.08 Preclinical evaluation of a small molecule inhibitor of WDR5 in facioscapulohumeral muscular dystrophy (FSHD)
Frida Karakashi, Ospedale San Raffaele

1:00–2:00 p.m.
LUNCH BREAK & POSTERS
LEO's International Flavors

SESSION 5: Clinical Care & Related Issues

Co-chairs: Nicol Voermans & Jeffrey Statland

2:00 p.m.
S5.01 Classification, clinical care, outcome measures, and biomarkers in childhood-onset facioscapulohumeral muscular dystrophy (FSHD): An update from the 279th European Neuromuscular Centre (ENMC) Workshop, the Netherlands, November 2024
Katy de Valle, Royal Children's Hospital

2:20 p.m.
S5.02 Focus on fatigue: The presence of performance fatigability in childhood-onset facioscapulohumeral dystrophy
Corrie Erasmus, Radboud University Medical Center

2:40 p.m.
S5.03 An updated international standard of care for facioscapulohumeral muscular dystrophy
Nicol Voermans, Radboud University Medical Center, & Raj Badiani, FSHD UK

3:00 p.m.
S5.04 Facioscapulohumeral dystrophy (FSHD): In search of a Brazilian epidemiological profile
Gabriella Dousseau, University of São Paulo

3:20 p.m.
S5.05 Addressing the unmet need for advance care planning and palliative care in FSHD
Emma Weatherley, FSHD Global Research Foundation, & Nicol Voermans, Radboud University Medical Center

3:40–4:00 p.m.
COFFEE BREAK & NETWORKING
Foyer

SESSION 6: Clinical Studies & Trial Design

Co-chairs: Nicol Voermans & Channa Hewamadduma

4:00 p.m.
S6.01 Introducing BetterLife FSHD, an innovative new patient-reported data repository and engagement tool
Amanda Hill, FSHD Society

4:20 p.m.
S6.02 Natural history of muscle volume and muscle fat content biomarkers in FSHD based on whole-body fat-referenced MRI
Markus Karlsson, AMRA Medical

4:40 p.m.
S6.03 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

5:00 p.m.
S6.04 Motor Outcomes to Validate Evaluations in Pediatric facioscapulohumeral muscular dystrophy (MOVE Peds): Protocol for an observational study
Michaela Walker, University of Kansas Medical Center

5:20 p.m.
S6.05 Avidity clinical trial interim data
Speaker TBA

5:40 p.m.
YOUNG INVESTIGATOR & BEST POSTER AWARDS
Conference program committee

5:50 p.m.
CLOSING REMARKS
Lucienne Ronco, Stephen Tapscott & Nicol Voermans

7:00–9:00 p.m.
JOINT IRC AND FSHD CONNECT RECEPTION

POSTERS

Amstel 3

The main poster session is on Thursday, June 12, 6:30-8:00 p.m. Poster presentations are on the following schedule: Odd-numbered posters 6:30-7:15 p.m., and even-numbered posters 7:15-8:00 p.m. The poster hall will also be open at lunch on both days of the meeting.

POPULATION GENETICS & MODIFIERS

P1.01 Genetic epidemiology of facioscapulohumeral muscular dystrophy in Hungary

Zsófia Flóra Nagy, Szabolcs Udvari, Xenia Jockers, Zoltán Grosz, Péter Balicza, Mária Judit Molnár

P1.02 Estrogen rescues muscle regeneration impaired by DUX4 in a humanized xenograft mouse model

Silvia Maiullari, Giada Mele, Patrizia Calandra, Giorgia di Blasio, Sonia Valentini, Alessio Torcinaro, Isabella Manni, Emanuela Teveroni, Fabio Maiullari, Maria Pesavento, Ludovica Giorgini, Sabrina Putti, Sara Bortolani, Enzo Ricci, Siro Luvisetto, Massimiliano Mazzone, Giancarlo Deidda, **Fabiola Moretti**

P1.03 Progesterone may be a regulator and B12 could be an indicator of the proximal D4Z4 repeat methylation status on 4q35ter

Ceren Hangül, Filiz Özcan, Sule Darbas, Hilmi Uysal, Ayse Filiz Koc, Sibel Berker Karauzum

P1.04 FSHD onset before 18 years of age: A retrospective longitudinal study

Beatrice Ravera, Carmine Di Marco, Ariele Barreto-Haagsma, Sara Bortolani, Eleonora Torchia, Mauro Monforte, Robert Muni-Lofra, Jordi Diaz-Manera, Michela Guglieri, Chiara Marini-Bettolo, Volker Straub, Enzo Ricci, Giorgio Tasca

P1.05 Beyond the classical definition: Unusual genetic patterns and clinical diversity in FSHD

Serpil Eraslan, Sahin Avci, Manar Kaptan, Ayse Nur Ozdag Acarli, Beyza Yavuzcan, Ilker Eren, Gulshan Yunisova, Mehmet Demirhan, Piraye Oflazer, Hulya Kayserili

MEASURES OF DISEASE ACTIVITY & PROGRESSION

P2.01 Whole-body muscle MRI in children: Two-year follow-up of the iFSHD-LOS cohort

Ian Woodcock, Markus Karlsson, Per Widholm, Michael Kean, Katy de Valle

P2.02 Electrical impedance myography captures features of muscle structure measured by MRI and transcriptomic analysis in facioscapulohumeral muscular dystrophy

Leo Wang, Buket Sonbas Cobb, Lara Riem, Olivia DuCharme, Dennis Shaw, Michaela Walker, Leann Lewis, Rabi Tawil, Johanna Hamel, Karlien Mul, Silvia Blemker, Katy Eichinger, Stephen Tapscott, Seward Rutkove, Jeffrey Statland

P2.03 Mapping immune dysregulation in FSHD: Toward the first immune atlas and novel therapeutic strategies

Andrea Papait, Lorena Di Pietro, Elsa Vertua, Eleonora Torchia, Sara Bortolani, Mauro Monforte, Elvira Ragozzino, Flavia Giacalone, Diego Sibilia, Elisa Orecchini, Andrea Sabino, Marta Barba, Angela Cozzolino, Clarissa Ferrari, Antonietta Rosa Silini, Enzo Ricci, Ornella Parolini

P2.04 Characterization of FSHD1 model mice carrying 5Mb chromosome 4q35 on mouse artificial chromosome

Yosuke Hiramuki, Miwa Hosokawa, Ichizo Nishino, Hiroyuki Kugoh, Yasuhiro Kazuki

P2.05 Quantitative whole-body MRI biomarker relation to muscle strength and function in FSHD patients

Mary Foltz, Per Widholm, Markus Karlsson, Jonathan Pini, Angela Puma, Luisa Villa, Michele Cavali, Andra Ezaru, Guillaume Bassez, Teresinha Evangelista, Romain Thomas, Loïc Danjoux, Céline Tard, Sabrina Sacconi

P2.06 Patient-derived stem cell models of childhood-onset facioscapulohumeral muscular dystrophy (FSHD) mirror disease severity *in vitro*

Peter Houweling, Vanessa Crossman, Kathrin Mattes, Leon Kiriaev, Chrystal Tiong, Natasha Tuano, Rebecca McElroy, Chantal Coles, Peter Jones, Richard Mills, Katy de Valle, Ian Woodcock

P2.07 Associations between muscle strength and MRI biomarkers in FSHD: Toward imaging-based functional classification

Per Widholm, Mary Foltz, Markus Karlsson, Jonathan Pini, Angela Puma, Luisa Villa, Michele Cavali, Andra Ezaru, Guillaume Bassez, Benjamin Marty, Teresinha Evangelista, Romain Thomas, Loïc Danjoux, Celine Tard, Sabrina Sacconi

P2.08 Expanding the UK FSHD Patient Registry Dataset: Improving data collection and amplifying "patient voice"

Helen Walker, Lucy Hickson, Robert Muni-Lofra, Emma Matthews, Rajeshri Badiani, Andrew Graham

P2.09 The UK Facioscapulohumeral Muscular Dystrophy Patient Registry: A powerful tool to support clinical research and patient voice in the translational research pathway

Lucy Hickson, Helen Walker, Chiara Marini-Bettolo, Robert Muni-Lofra, Andrew Graham, Fiona Norwood, Mark Roberts, Tracey Willis, Emma Matthews, Mark McQueen-Mencias, Giorgio Tasca, Gavin Langlands, Grace McMacken, Kate Adcock

P2.10 Linking mechanical strain to progression of fat infiltration in FSHD

Kimberly Steininger, Allison McCrady, Seth Friedman, Stephen Tapscott, Silvia Blemker

NOVEL CLINICAL OUTCOME MEASURES

P3.01 Quantitative muscle ultrasound as a clinical correlate in facioscapulohumeral muscular dystrophy: Validation of a rapid assessment protocol

Nurit Birman Har-Noy, Omer Bouzaglo, Vivian E. Drory, Alon Abraham

P3.02 Quantitative muscle ultrasound as a clinical correlate in facioscapulohumeral muscular dystrophy: Validation of a rapid assessment protocol

Odette van Iersel, Harm Weekenstroo, Donnie Cameron, Jonne Doorduin, Nens van Alfen

P3.03 Measuring upper and lower limb movement with Syde® in patients with facioscapulohumeral muscular dystrophy (FSHD): Analytical validation in a controlled environment

Peter Ankjær, Mads G Stemmerik, Bjørk Teitsdòttir, Anna Slipsager, Laura Desire, Nicolas Noblot, Oihana Piquet, Tammy McIver, Paul Strijbos, Stéphane Motola, **Alexis Tricot**, John Vissing, Nicolai Preisler

P3.04 Episodes of symptoms worsening in FSHD patients

Nurit Birman Har-Noy, Jacopo Luca Casiraghi, Valeria Ada Sansone

MECHANISMS OF DISEASE & INTERVENTIONAL STRATEGIES

P4.01 Quantifying anti-DUX4 therapy for facioscapulohumeral muscular dystrophy

Matthew V. Cowley, Peter S. Zammit, Christopher R. S. Banerji

P4.02 Characterization of a DUX4-responsive reporter mouse

Lindsay Wallace, Jessica Camp, Kate Neal, Noah Taylor, Scott Harper

P4.03 Investigating Dux expression in extra-embryonic tissues and whether it modulates maternal-fetal tolerance

Aidan O'Donnell, Andrew Smith, Sean Bennett, Stephen Tapscott

P4.04 Modeling cell-type specific sporadic DUX4 activation in FSHD

Mitsuru Sasaki-Honda, Alvaro Rada-Iglesias, Hidetoshi Sakurai

P4.05 SMCHD1 enzymatic activity is promoted by bidentate binding to DNA

Alexandra Gurzau, Richard Birkinshaw, Andrew Leis, James Murphy, Marnie Blewitt

P4.06 Detailed analysis of inflammatory and ultrastructural changes in TIRM-MRI guided muscle biopsies of FSHD patients

Anna Greco, Benno Kusters, Ritse Mann, Jurgen Fütterer, Leon de Jong, Yordy Welling, Marieke Ploegmakers, Ger Pruijn, Leo Joosten, Baziel van Engelen

P4.07 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, Anne-Emilie Declèves, Alexandra Belayew, Frédérique Coppée, Alexandra Tassin

P4.08 DUX4-sPAS base editing gene therapy technology in FSHD
V. Dauksaite, D. Vinke, R. Augustinus, D. Zheng, J. M. Janssen, J. Liu, M. F. V. Gonçalves, J. C. de Greef, N. Geijsen, S. M. van de Maarel

P4.09 Unlocking the potential of 3D-TESMs: A promising drug testing tool for FSHD therapies
Silvana Jirka, Lucia Bartolotti, Marnix Franken, Silvère van der Maarel, Jessica de Greef

P4.10 Increased METTL3 expression and m6A methylation in myoblasts of facioscapulohumeral muscular dystrophy
Nikolaos Settas, Adam Bittel, Yi-Wen Chen

P4.11 DUX4 overexpression in proliferating myoblasts induces an early response of the metabolic sensor AMPK
Fiona Ngassam Lowe, Charlotte Lemoine, Maëlle Sciot, Alexandre Legrand, Professor Alexandra Tassin, Anne-Emilie Decleves

P4.12 Application of the Cuore to analyze effect of training on FSHD 3D tissue engineered skeletal muscles
Galina Filonova, Marnix Franken, W. W. M. Pim Pijnappel, Silvana M. G. Jirka, Silvère M. van der Maarel, Jessica C. de Greef

P4.13 Baseline expression of DUX4-regulated pathogenic genes across 74 different human muscles in healthy controls
Aysylu Murtazina, Artem Borovikov, Anna Kuchina, Daria Sherstukova, Mikhail Skoblov

P4.14 Fibro-adipogenic progenitors and FSHD
Carlo Serra, Kathryn Wagner, Thomas Lloyd

P4.15 A discrete region of the D4Z4 is sufficient to initiate epigenetic silencing
Ellen Paatela, **Faith St. Amant**, Danielle Hamm, Sean Bennett, Taranjit Gujral, Silvère van der Maarel, Stephen Tapscott

P4.16 Targeting DUX4 transcriptional activity with engineered DNA-binding repressors: A novel therapeutic approach for FSHD
Heloise Hoffmann, Alice Finkelstein, Goldie Roth, Michael Liu, Vanessa Chiprez Meza, Amanuel Geremew, Ayushi Mohanty, Katherine Xu, Maria Fernanda Velásquez, Phillip Kyriakakis, Lei S. Qi

P4.17 Direct RNA sequencing reveals the altered epitranscriptomic landscape in DUX4-expressing myoblasts

Dongxu Zheng, Judit Balog, Iris M. Willemsen, Anita van den Heuvel, Hailiang Mei, Susan Kloet, Ahmed Mahfouz, Silvère M. van der Maarel

P4.18 An update: Creating an immune cell atlas of the peripheral blood for facioscapulohumeral muscular dystrophy (FSHD)

Chantal Coles, Katy de Valle, Ian Woodcock, **Peter Houweling**

P4.19 Targeting DUX4 mRNA with anti-TfR NANOBODY® oligonucleotide conjugates

Elizabeth Allen, Fernande Freyermuth, Wen Tang, Samantha Gentle, Jayaprakash Thummapudi, Shan Zhou, Shruthi Ramkumar, Ayman Ismail, Sigrid Cornelius, Nina Leksa

P4.20 Non-myogenic mesenchymal cells shape the degenerative microenvironment in FSHD patient muscles

Flavia Giacalone, Lorena Di Pietro, Alessandra Nagar, Elisa Orecchini, Chiara Lisciandrello, Gaia Guardabascio, Simona Nanni, Giulia Mantini, Andrea Papait, Elvira Ragozzino, Diego Sibilia, Federica Nagar, Sara Bortolani, Eleonora Torchia, Andrea Sabino, Wanda Lattanzi, Luciano Giacò, Enzo Ricci, Ornella Parolini

P4.21 Nucleolar FRG2 lncRNAs inhibit rRNA transcription and translation linking FSHD to dysregulation of muscle-specific protein synthesis

Valentina Salsi, **Francesca Losi**, Bruno Fosso, Marco Ferrarini, Sara Pini, Marcello Manfredi, Gaetano Vattemi, Tiziana Mongini, Lorenzo Maggi, Graziano Pesole, Anthony Henras, Paul Kaufman, Rossella Tupler

P4.22 The overexpression of the RNA binding protein FRG1 leads to reduced maturation and decreased metabolic efficiency in skeletal muscle

Sebastian Fantini, Grazia Bisceglia, Joanna Zyla, Noemi Moio, Andi Nuredini, Giuseppe D'Antona, Gaetano Vattemi, Edoardo Malfatti, Joanna Polanska, **Rossella Tupler**

P4.23 DUX4 as a co-regulator of hormone nuclear receptors in human myoblasts

Camila Simonetti, Sabrina M. Pagnoni, Oliver D. King, Scott Q. Harper, **Alberto L. Rosa**

P4.24 Role of miR-200c and oxidative stress in FSHD

Francesca De Santa, Valentina Fustaino, Giorgia Sperandio, Giada Mele, Alessandra Magenta, Ferdinando Scavizzi, Anne Bigot, Vincent Mouly, Fabiola Moretti, **Alessio Torcinaro**

P4.25 Structural basis for the interactions of DUX4 with Med25 and CBP/p300
Moriya Slavin, **Clothilde Claus**, Keren Zohar, Karimatou Bah, Tsiona Eliyahu, Michal Linial, **Frédérique Coppée**, **Nir Kalisman**

P4.26 Evidence for the protective effects of estrogens in a mouse model of FSHD
Adam Bittel, Yi-Wen Chen

P4.27 Non-viral generation of patient-derived iPSCs for modeling FSHD and screening nanoparticle-based therapeutics
Deepali Shukla, Dinesh Babu, R. Velayudhan Shaji, Meenal Kowshik, Indrani Talukdar

CLINICAL CARE & RELATED ISSUES

P5.01 Neuropsychological profiles of children and young people with childhood-onset facioscapulohumeral dystrophy (FSHD)
Louise Crowe, Ian Woodcock, Chelsea Finchett, Vicki Anderson, **Katy de Valle**

P5.02 FSHD UK: From voluntary group to a registered charity creating a strong multi-stakeholder group to strategically drive clinical trial readiness and co-ordination of FSHD activities in the UK
Rajeshri Badiani

P5.03 Strengthening regional neuromuscular care in the Netherlands: Preparing for future FSHD trials and care demands
Nicole Voet, Martin Beuzel, Kirsten IJsebaert, Anita Beelen, Esther Kruitwagen, Charlotte van Esch

P5.04 Deviating observations when comparing D4Z4 FSHD repeat analysis on the Bionano OGM platform with the linear Southern blot in FSHD diagnostics
Saskia Smith, Anneliese Grimbergen, Thomas Wijnands, Dennis Verbove, **Nienke van der Stoep**

P5.05 The True Cost of FSHD: A burden of illness study of facioscapulohumeral muscular dystrophy patients in the United States
Amanda Hill, June Kinoshita, Marina Kolocha, **Man Hung**, Eric Hon, **Jamshid Arjomand**

P5.06 Project Mercury: A global platform for accelerating therapeutic development and patient access in FSHD

Amanda Hill, Miriam Wagner Long, Josie Godfrey, June Kinoshita, Anna Gilmore, Neil Camarta, Mark Stone, Ken Kahtava

P5.07 AI-driven innovation in FSHD: Next-generation support for patients and providers?

Claudia Strafella, Domenica Megalizzi, Giulia Trastulli, Emma Proietti Piorgo, Luca Colantoni, Francesca Torri, Barbara Risi, Carlo Caltagirone, Giulia Ricci, Gabriele Siciliano, Massimiliano Filosto, Raffaella Cascella, Emiliano Giardina

P5.08 Co-occurrence of anti-AChR myasthenia gravis in facioscapulohumeral dystrophy patients: A case series

Giulia Tammam, Luisa Villa, Richard JLF Lemmers, Jonathan Pini, Leonardo Salviati, Michele Cavalli, Andra Ezaru, Angela Puma, Silvère M. van der Maarel, **Sabrina Sacconi**

P5.09 Starting the Italian Registry for Facioscapulohumeral Muscular Dystrophy

Giulia Ricci, Barbara Risi, Francesca Torri, Vercelli Liliana, Chiara Colombi, Emiliano Giardina, Claudia Strafella, Angela Berardinelli, Gabriele Siciliano, Tiziana Mongini, Massimiliano Filosto

P5.10 Late-onset facioscapulohumeral muscular dystrophy as a differential diagnosis issue

Ayse Nur Ozdag Acarli, Fahrettin Sertac Yapar, Gulshan Yunisova, Serpil Eraslan, Sahin Avci, Ilker Eren, Mehmet Demirhan, Hulya Kayserili, Piraye Oflazer

P5.11 Development of a disease progression model for FSHD to support health technology assessment

Margaret Petrou, Nicholas Brighton, Amanda Hill, **Josie Godfrey**

P5.12 Sexual health and pelvic floor function in women with facioscapulohumeral dystrophy

Brianna Brun, Jeanne Dekdebrun, Samia Lopa, Erin Richardson, Loralei Thornburg, Martina Anto-Ocrah, Johanna Hamel

CLINICAL STUDIES & TRIAL DESIGNS

P6.01 Making strength training work in FSHD

Ronne van Haaren-Pater, Nicole Voet, Mariska Janssen, Nicol Voermans, Ton Satink

P6.02 Improving routine care for facioscapulohumeral muscular dystrophy: Effectiveness of personalised antioxidant supplementation on muscle strength and quality of life in a real-world setting

Sandrine Arbogast, Dalila Laoudj-Chenivesse, Eric Raynaud De Mauverger, Christine Fedou, Emma Debroize, Gérald Hugon, Joël Pincemail, Marie-Christine Picot, Jean-Paul Cristol, Jacques Mercier, Florence Portet

P6.03 A toolkit for new facioscapulohumeral muscular dystrophy trial sites

Joost Kools, Lawrence Korngut, Janet Ballantyne, Irene Roozen, Ria de Haas, Amanda Hill, Teresinha Evangelista, Valeria Sansone, Richard Roxburgh, Hanns Lochmuller, Jeff Statland, Nicholas Johnson, Nicol Voermans

P6.04 The FOCUS 3 study protocol: Ten-year follow-up in facioscapulohumeral dystrophy

Eline Boon, Sanne Vincenten, Jildou Dijkstra, Corrie Erasmus, Nicole Voet, Karlien Mul, Nicol Voermans

P6.05 Optimising clinical development programs for facioscapulohumeral muscular dystrophy (FSHD) therapies in preparation for Joint Clinical Assessment (JCA) in Europe

Johann Sanseau, **Sophie Schmitz**

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, Corrie Erasmus, Tracey Willis, Nicol Voermans

P6.07 Comorbidities, medication use, and adverse events in FSHD patients: Insight from the ReSOLVE and MOVE studies

Iwona Skorupinska, Enrico Bugiardini, Louise Germain, Jose M Sanz Mengibar, Laura Berg, Anna Ubiali, Russell Butterfield, Elena Carraro, John Day, Nuran Dilek, Stacy Dixon, Bakri Elsheikh, Katy Eichinger, Seth Friedman, Channa Hewamadduma, Kiley Higgs, Nicholas Johnson, Leann Lewis, Doris G Leung, Hanns Lochmuller, William B Martens, Michael McDermott, Karlien Mul, Sabrina Sacconi, Valeria Sansone, Dennis Shaw, Perry Shieh, Sub Subramony, Jaya Trivedi, Kathryn Wagner, Michaela Walker, Leo H Wang, Matthew Wicklund, Rabi Tawil, Jeff Statland, all investigators of FSHD CTRN NA

P6.08 A hub-and-spoke model for the French National FSHD Registry: A 2025 update

Benoît Sanson, Céline Guien, Sitraka Rabarimeriarijaona, Rafaëlle Bernard, Christophe Béroud, Sabrina Sacconi, the French FSHD registry collaboration group

P6.09 Early-onset FSHD: An Italian case series

Martina Prestamburgo, Claudia Conti, Arianna Iosca, Laura Carraro, Irene Dainesi, Selvia Khalil, Alice Gardani, Lorenzo Maggi, Giulia D'alvano, Federica Silvia Ricci, Rossella D'alessandro, Andrea Barp, Massimiliano Filosto, Liliana Vercelli, Giulio Gadaleta, Tiziana Enrica Mongini, Mauro Monforte, Beatrice Ravera, Giulia Ricci, Lucia Ruggiero, Grazia d'Angelo, Emanuele Costantini, Lorenzo Verriello, Guja Astrea, Roberta Battini, Stefano Previtali, Antonella Pini, Chiara Fiorillo, Angela Berardinelli

P6.10 The landscape of FSHD data collection: A 2025 expansion to the TREAT-NMD FSHD dataset

Farjana Ali, Mitsuru Sasaki-Honda, Helen Walker, Richard Roxburgh, Raj Badiani, Dino Masic, Annie Poll, Miriam Rodrigues, Michaela Guglieri, Anna Ambrosini

P6.11 The natural history of childhood-onset FSHD in an Australian cohort: iFSHD-LOS 2-year data

Katy de Valle, Chantal Coles, Peter Houweling, Ian Woodcock

LATE-BREAKING ABSTRACTS

Will be added after May 17