32nd International
Research Congress
on FSHD

Powering THE PATH FORWARD

JUNE 12-13, 2025 | LEONARDO ROYAL HOTEL | AMSTERDAM, THE NETHERLANDS

ABSTRACTS HERE

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HARTELIJK WELKOM IN *Amsterdam!*

Welcome to Amsterdam and the 32nd Annual International Research Congress on FSHD. Set in a city long celebrated for its contributions to art, innovation, and discovery – from the Dutch Golden Age to the birth of microbiology – Amsterdam provides an inspiring backdrop for this year's gathering. It's a fitting place – where history and progress converge – to explore bold new directions in FSHD research.

This year's scientific program reflects the extraordinary momentum across key domains of FSHD research. Presentations span the validation of clinical outcome measures in both pediatric and adult populations through the MOVE studies, integration of whole-body MRI with molecular biomarkers to map disease progression, and the emergence of platforms like BetterLife FSHD to elevate patient engagement. Insights into comorbidities and treatment responses, alongside expanded trial networks across Europe, Australia, Brazil, Hong Kong, and Japan, speak to a field increasingly focused on clinical readiness and global collaboration. These efforts underscore a shared goal: Building robust, reproducible, and patient-centered paths to therapeutic development.

Together, we're charting a future where meaningful interventions for FSHD are not only possible, but within reach. Your work, your presence, and your commitment make this progress real – and we're honored to continue the journey with you here in Amsterdam.

PROGRAM COMMITTEE

- Lucienne Ronco, PhD, organizing chair
- Nicol Voermans, MD PhD, co-chair
- Stephen Tapscott, MD PhD, co-chair
- Marnie Blewitt, PhD
- Enrico Bugiardini, PhD
- Seth Friedman, PhD
- Channa Hewamadduma, FRCP
- Cristiane Araujo Martins Moreno, MD PhD
- Jeffrey Statland, MD
- Giorgio Tasca, MD PhD
- Kyoko Yokomori, PhD
- Vishnu VY, MD

KEYNOTE SPEAKERS

Marion Sellenet (on film) is a 35-year-old visual artist from the Cévennes who lives in Brussels. Diagnosed at age 15 with FSHD, she says: "I made my film, *Marion and the Metamorphosis*, to cure myself of a second illness, not FSHD per se, but one I developed when I was diagnosed: That of the sword of Damocles, of fear, despair, and shame. Because where medicine is powerless to make symptoms disappear, it has no tools to offer for living well with the condition. I made this film to pick up the pieces of my being, fractured by figures and words I didn't understand and by a negative societal vision of the disease. Paradoxically, it was by giving up all hope of a cure that I regained hope in my life. I aspire now to a collaboration among patients, doctors, researchers, and philosophers, with the aim not just of curing symptoms, but of curing people and developing knowledge of how to live healthily with illness."

Leendert Trouw, PhD, is a senior researcher and professor of immunology at Leiden University Medical Center (LUMC), where he focuses on the role of complement and autoantibodies in autoimmune disease. He was the first to demonstrate the pathogenicity of anti-C1q autoantibodies during his doctoral studies, and his postdoctoral work in Sweden highlighted the role of complement inhibitors in binding free DNA and dead cells. At LUMC, Dr. Trouw's team discovered anti-carbamylated protein (anti-CarP) antibodies in rheumatoid arthritis, which have since been shown to predict disease onset and severity and are now being developed into a commercial diagnostic test. Supported by prestigious Dutch grants (Veni, Vidi, and the European Research Council), his research continues to explore how modified self-proteins drive autoantibody responses and how complement contributes to disease progression. Recently, he transitioned his group to the Department of Immuno-Hematology to broaden the impact of his findings across multiple disease areas.

Mart Brandjes was diagnosed with FSHD at age 14. Since then, his physical abilities have gradually declined. He now relies on mobility aids, and many everyday activities are no longer possible. Between 2019 and 2024, Brandjes participated in a clinical drug trial and several other FSHD-related research studies. Since 2022, he has been involved with SingelSwim, a fundraising initiative for FSHD research, through which he connected with the FSHD Stichting, a foundation based in the Netherlands. In 2024, he joined the foundation's board. He has been working in the yacht-building industry since 2018, where he designs, calculates, and guides complex engineering projects through to successful delivery.

Karen Chen, PhD, is the chief executive officer of the Spinal Muscular Atrophy (SMA) Foundation, where she oversees scientific strategy, drug discovery, and operational activities. With more than 35 years of experience in neuroscience research and management, she has led preclinical programs in both nonprofit and pharmaceutical settings. Before joining the SMA Foundation, Dr. Chen held leadership roles at Roche Palo Alto and Elan Pharmaceuticals, focusing on therapeutics for Alzheimer's, Parkinson's, and other neurodegenerative diseases. She completed her postdoctoral training at Genentech and earned her PhD in neuroscience from the University of California, San Diego, working with Dr. Fred "Rusty" Gage. Dr. Chen has authored more than 50 scientific publications, holds multiple patents, and actively serves on several nonprofit boards, including Solve FSHD and the Bluefield Project. She is also CEO and cofounder of Imago Pharmaceuticals and previously served on the National Advisory Neurological Disorders and Stroke Council.

SLIDE PRESENTATIONS: 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

SLIDE PRESENTATIONS: DAY 1

Thursday, June 12, 2025 • AMSTEL 1 & 2

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.
SPOTLIGHT FIVE-MINUTE POSTER TALKS

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

EVENING SESSION

6:00–6:30 p.m.
SPOTLIGHT FIVE-MINUTE POSTER TALKS
Amstel 1 & 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

SLIDE PRESENTATIONS: 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, researchers are developing approaches to improve muscle function that could be used in combination with the approved treatments. 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
Nikolaos Settas presenting for Adam Bittel, Children's National Hospital

SLIDE PRESENTATIONS: DAY 2

Friday, June 13, 2025 • AMSTEL 1 & 2

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
Jildou Dijkstra, 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 Topline data from dose escalation cohorts A and B in FORTITUDE™, a phase 1/2 trial evaluating Del-brax (delpacibart braxlosiran) in adults with facioscapulohumeral muscular dystrophy (FSHD)
Jeffrey Statland, University of Kansas Medical Center

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

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

6:00 p.m.
BREAK & ADJOURN

EVENING SOCIAL TIME

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

POSTER PRESENTATIONS • 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 See P6.12

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 Exploring contractile performance with ultrasound in FSHD: Preliminary results of the MUSCLE+ Study
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, Alexandra Tassin, Anne-Emilie Declèves

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, C. Hunter Wallace, Kristen Peissig, Wen Tang, Samantha Gentle, Jayaprakash Thummapudi, Shan (Julia) Zhou, Shruthi Ramkumar, Ayman Ismail, Sigrid Cornelis, Kevin J. Kim, Anthony Saleh, Nina Leksa, Fernande Freyermuth

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

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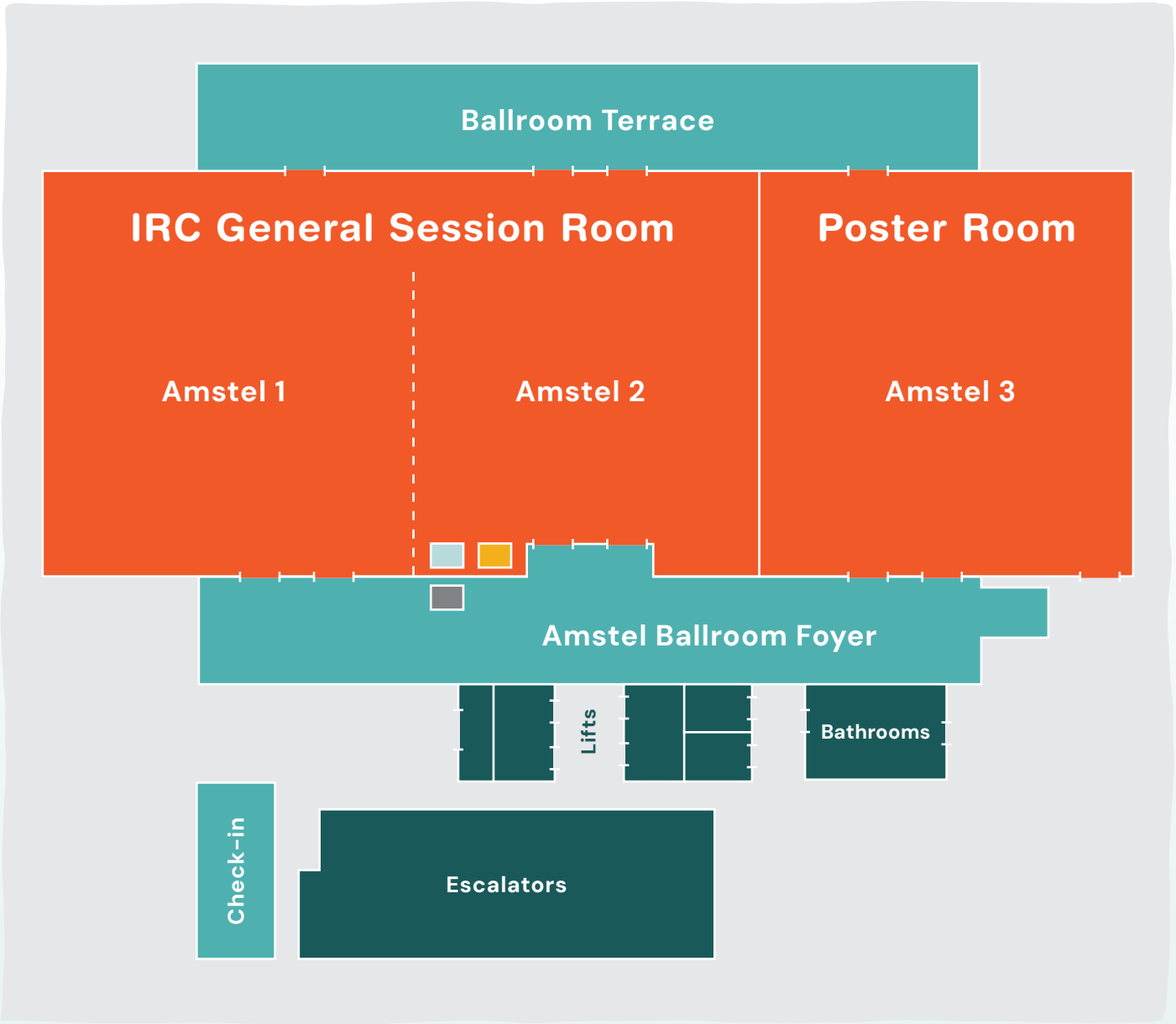

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- Researchers search and interact with patient data using the BetterLife FSHD Research Gateway

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