29th International Research Congress on FSHD

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

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We would like to welcome you to the FSHD Society’s 29th annual International Research Congress. This meeting, long established as the premier global platform for the discussion and dissemination of the state-of-the-art basic and clinical research on facioscapulohumeral muscular dystrophy (FSHD), is once again convening in-person, at the beautiful Waldorf Astoria Bonnet Creek hotel in Orlando, Florida. There will be a virtual simulcast of the meeting for those who may not be able to attend the congress in person.

For the past two years, we have greatly appreciated your willingness to participate in and attend the IRC virtually. During that time, despite the extended disruptions, the research community persevered and continued to make great strides in advancing the field and our understanding of FSHD pathophysiology. It was also exciting to see that clinical programs were able to adapt and continue to provide care for the patient community.

In addition, we were delighted to see a growing number of participants, as well as a broader international representation, participate in these meetings. The opportunity to hold virtual conferences made us aware that travel is not always possible for many dedicated researchers, and that wider dissemination of cutting-edge research can only help accelerate the development of treatments for the global patient community. To that end, just as we committed to alternate the congress between the United States and Europe as a way of expanding accessibility, the 2022 IRC and subsequent meetings will be planned as hybrid events, allowing broader representation and participation of the global research community.

For the 2022 IRC, we look forward to having the world’s foremost clinicians, medical researchers, pharmaceutical industry leaders, and basic scientists present and discuss new developments, reinforce collaborative efforts, facilitate new initiatives, and coordinate research and clinical activities. With the recent progress in FSHD research and clinical advances, this conference has become catalytic in translating ideas into potential therapies, and we look forward to this year’s program and your participation.

Program Committee

Jamshid Arjomand, PhD (organizer)
Julie Dumonceaux, PhD (co-chair)
Doris Leung, MD PhD (co-chair)
Russell Butterfield, MD PhD
Amy Campbell, PhD
Yi-Wen Chen, DVM PhD

Mikell Lang (program manager)
Frédérique Magdinier, PhD
Valeria Sansone, MD PhD
Giorgio Tasca, MD PhD
Yegor Vassetzky, PhD
Lindsay Wallace, PhD
DAY 1 – THURSDAY, JUNE 16, 2022

SLIDE PRESENTATIONS
Grand Ballroom

8:00 a.m.
WELCOMING REMARKS

8:15 a.m.
KEYNOTE: Patient Perspective on Living with FSHD
Lexi Pappas

8:30 a.m.
KEYNOTE: Facts and Fiction: How DUX4 Came on Stage, Could Play Good or Bad Guy, and Might Still Surprise Us
Alexandra Belayew, MSc PhD, University of Mons

9:30–11:10 a.m.
SESSION 1: Discovery Research
Session Chairs: Yi-Wen Chen, DVM PhD, & Julie Dumonceaux, PhD

9:35 a.m.
S1:101—Transcriptional and Post-transcriptional Mechanisms Induced by DUX4 Reshape Protein Synthesis
Danielle Hamm

9:55 a.m.
MORNING NETWORKING BREAK

10:30 a.m.
S1:102—DUX4 Expression Activates JNK and p38 MAP Kinases in Myoblasts
Nicolas Christoforou, Christopher Brennan, Abby Hill, Vijaya Madeti, Susanne Breitkopf, Seth Garren, Liang Xue, Tamara Gilbert, Angela Hadjipanayis, Maria Monetti, Charles P. Emerson J r., Rob Moccia, James Owens

10:50 a.m.
S1:103—Structure-Function Characterization of a DUX4 Inhibitor for a Drug-Like Approach to Treat FSHD Muscular Dystrophy
Paola Ghezzi, Andrea Berardi, Valeria Runfola, Maria Pannese, Giovanna Musco, Davide Gabellini

11:10 a.m.–12:15 p.m.
SESSION 2: Genetics & Epigenetics
Session Chairs: Frédérique Magdinier, PhD, & Russell Butterfield, MD PhD

11:15 a.m.
S2:201—A Novel Epigenetic Activator of DUX4 for the Therapy of FSHD Muscular Dystrophy
Emanuele Mocciaro, Roberto Giambruno, Stefano Micheloni, Maria Pannese, Valeria Runfola, Giulia Ferri, Davide Gabellini

11:35 a.m.
S2:202—dCAS-CTCF Modifies 3D Genome Organization and DUX4 Expression in FSHD Myoblasts
Anna Karpukhina, Evgenia Tuikacheva, Zhenru Pan, Sergey Ulyanov, Yegor Vassetzky

11:55 a.m.
S2:203—Nanopore Sequencing Reveals Size-Dependent Methylation Gradients in D4Z4 Repeat Arrays
Russell Butterfield, Diane M Dunn, Brett O Duvall, Sarah Moldt, Brith Otterud, Kristen Wong, Robert B Weiss

12:15 p.m.
LUNCH

1:00–3:00 p.m.
POSTER VIEWING & NETWORKING
(ODD NUMBERS)
Floridian Ballroom Foyer (see map on page 11)

3:00–4:25 p.m.
SESSION 3: Pathology & Disease Mechanisms
Session Chairs: Amy Campbell, PhD, & Yegor Vassetzky, PhD

3:05 p.m.
S3:301—DUX4 RNA G-quadruplexes as Drivers of Cellular RNA-Protein Granule Formation in FSHD
Prakash Kharel, Paul Anderson, Pavel Ivanov

3:25 p.m.
S3:302—DUX4 Activation of Human Pericentric Satellite Repeats Impairs DNA Damage Signaling
Tessa Arends
3:45 p.m.
S3:303—Interplay Between Mitochondrial Reactive Oxygen Species, Oxidative Stress and Hypoxic Adaptation in FSHD: Metabolic Stress as Potential Therapeutic Target
**Philipp Heher, Massimo Ganassi, Adelheid Weidinger, Elise Engquist, Johanna Pruller, Thuy Hang Nguyen, Alexandra Tassin, Anne-Emilie Decleves, Kamel Marchaoui, Christopher Banerji, Johannes Grillari, Andrey Kozlov, Peter Zammit**

4:05 p.m.
S3:304—Antiapoptotic Protein FAIM2 is a Regulatory Node, Downstream of DUX4-TRIM21 and miR-3202
**Michael Kyba, Hossam Soliman, Erik Toso, Inas Darwish, Samia Ali**

4:25 p.m.
DAY 1 CLOSING REMARKS
Jamshid Arjomand, PhD, FSHD Society

6:00 p.m.
RECEPTION
Golf Pavilion (#36 on map)

7:00 p.m.
BANQUET
Golf Pavilion (#36 on map)
11:20 a.m.
KEYNOTE: Data Aggregation and Disease Modeling to Accelerate Rare Disease Drug Development
Jane Larkindale, PhD, PepGen

12:00 p.m.
LUNCH

1:00–3:00 p.m.
POSTER VIEWING & NETWORKING (EVEN NUMBERS)
Floridian Ballroom Foyer (see map on page 11)

3:00–4:25 p.m.
SESSION 5: Clinical Studies & Outcome Measures
Session Chairs: Doris Leung, MD PhD, & Giorgio Tasca, MD PhD

3:05 p.m.
S5:501—Clinical Trial Readiness to Solve Barriers to Drug Development in FSHD (ReSolve): Baseline Characteristics
Jeffrey Statland, Kate Eichinger, Michael McDermott, Kiley Higgs, Michaela Walker, Doris Leung, Sabrina Sacconi, Karien Mul, Valeria Sansone, Elena Carraro, Leo Wang, Perry Shieh, Bakri Elsheikh, Samantha LoRusso, Russell Butterfield, Nicholas Johnson, Rabi Tawil, the ReSolve Investigators of the FSHD CTRN

3:25 p.m.
S5:502—Muscle Ultrasound as Imaging Biomarker in Facioscapulohumeral Muscular Dystrophy: Possibilities and Pitfalls
Sjan Teeselink, Sanne Vincenten, Nicol Voermans, Baziel van Engelen, Karlien Mul, Nens van Alfen

3:45 p.m.
S5:503—Diagnostic MRI Biomarkers for FSHD Identified by Machine Learning
Mauro Monforte, Sara Bortolani, Eleonora Torchia, Lara Cristiano, Francesco Laschena, Tommaso Tartaglione, Enzo Ricci, Giorgio Tasca

4:05 p.m.
S5:504—Reachable Workspace to Evaluate Efficacy of Losmapimod in FSHD in Two Phase 2 Studies
Christopher Morabito, J ay Han, Anthony Accorsi, J ordi Diaz, Miriam Freimer, Angela Genge, Summer Gibson, Nuria Gomez, Namita Goyal, J ohanna Hamel, Lawrence Hayward, J ohn J iang, Nicholas J ohnson, J oost Kools, David Reyes Leiva, Doris Leung, Hanns Lochmuller, Samantha Lorusso, Michelle L. Mellion, Alan Pestrnak, L. Alejandro Rojas, Sabrina Sacconi, Perry Shieh, J ennifer Shoskes, J effrey Statland, S.H. Subramony, Rabi Tawil, Baziel van Engelen, J uan Vilchez, Kathyrn Wagner, Leo Wang

4:25 p.m.
Best Poster Prize & Young Investigator Award
Moderators: Julie Dumonceaux, PhD, & Doris Leung, MD PhD

4:45 p.m.
2023 IRC ANNOUNCEMENT & FINAL REMARKS
Jamshid Arjomand, PhD, FSHD Society

4:50 p.m.
ADJOURN
Floridian Ballroom Foyer

**Odd-numbered presentations on Thursday, 1:00-3:00 p.m. Even-numbered presentations on Friday, 1:00-3:00 p.m.**

### DISCOVERY RESEARCH

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<td>Single cell sequencing identifies unique transcriptional responses to plasma membrane injury in FSHD</td>
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<td>P1.02</td>
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<td>Jessica de Greef, Linde Bouwman, Bianca den Hamer, Silvère Van der Maarel</td>
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### GENETICS & EPIGENETICS

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Yosuke Hiramuki, Ichizo Nishino, Hiroyuki Kugoh, Yasuhiro Kazuki

P3.17 Transcriptomic analysis of inflamed and non-inflamed FSHD muscle, together with peripheral blood mononucleated cells, reveals a circulating biomarker of clinical severity in FSHD
Christopher Banerji, Anna Greco, Leo J oosten, Baziel van Engelen, Peter Zammit

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P3.20 DUX4, nucleolar stress, and FSHD myopathy
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P3.21 Innate immunity model of FSHD muscle pathology activates complement genes
Katelyn Daman, Jing Yan, Oliver King, Michael Brehm, Charles P. Emerson Jr.

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P4.28 Identification and targeting of hypoxia signaling for translational potential in facioscapulohumeral muscular dystrophy
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Serpil Eraslan, Sahin Avcı, Ilker Eren, Gulshan Yunisova, Piraye Ofazer, Mehmet Demirhan, Hulya Kayerli

P5.52 Prevalence and disease progression of genetically confirmed facioscapulohumeral muscular dystrophy type 1 (FSHD1) in China between 2001 and 2020: a nationwide population-based study
Zhiqiang Wang, Liangliang Qiu, Minting Lin, Long Chen, Fuze Zheng

P5.53 Association between D4Z4 hypomethylation and disease severity: a retrospective cohort study in China
Zhiqiang Wang, Fuze Zheng, Long Chen, Liangliang Qiu, Lin Lin, Minting Lin, Ying Fu, Ning Wang
RESTAURANTS & LOUNGES

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15. Promenade
16. Signature Island
17. Signia by Hilton Meeting Rooms
18. Floridian Ballroom
19. Bonnet Creek Ballroom

AMENITIES & TRANSPORTATION

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