



29<sup>TH</sup> ANNUAL FSHD SOCIETY  
**INTERNATIONAL  
RESEARCH  
CONGRESS**

June 16–17, 2022  
Waldorf Astoria and  
Hilton Bonnet Creek Hotel  
Orlando, Florida



# 29<sup>th</sup> 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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# WELCOME, EVERYONE!

**W**e 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



# AGENDA

## 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 Bretkopf, Seth Garren, Liang Xue, Tamara Gilbert, Angela Hadjipanayis, Mara Monetti, Charles P. Emerson Jr., Rob Moccia, Jane 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, Zhenrui 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**





# AGENDA

**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 Mamchaoui, 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)**

**DAY 2 – FRIDAY, JUNE 17, 2022**

**SLIDE PRESENTATIONS**

**Grand Ballroom**

**9:00 a.m.**

**WELCOMING REMARKS**

**9:05 a.m.**

**KEYNOTE: Solve FSHD: Catalyst for a Cure for FSHD**  
**Eva Chin, PhD, Solve FSHD**

**9:25–11:20 a.m.**

**SESSION 4: Interventional Strategies**

**Session Chairs: Lindsay Wallace, PhD, & Valeria Sansone, MD PhD**

**9:30 a.m.**

**S4:401—Investigation of Human Bone Marrow Mesenchymal Stem Cell-Derived Extracellular Vesicles as Therapeutic Agents for Facioscapulohumeral Muscular Dystrophy (FSHD)**  
**Lindsay Wallace, Scott Harper, Nizar Saad**

**9:50 a.m.**

**S4:402—AOC 1020: An Antibody Oligonucleotide Conjugate (AOC) in Development for the Treatment of FSHD**

**Barbora Malecova**, David Sala, Garineh Mary Melikian, Guln Erdogan, Rachel Johns, Joanne Young, Erwann Ventre, Sole Gatto, Matthew Onorato, Orsolya Kiraly, Martin Kogler, Philipp Hadwiger, Lukas Perkams, Arthur Levin, Michael Flanagan

**10:10 a.m.**

**S4:403—AAV-CRISPR-Cas13 Gene Therapy for FSHD: DUX4 Gene Silencing Efficacy and Immune Responses to Cas13b Protein**

**Afroz Rashnonejad**, Gholamhossein Amini Chermahini, Noah Taylor, Allison Fowler, Emma Kraus, Oliver King, Scott Harper

**10:30 a.m.**

**MORNING NETWORKING BREAK**

**11:00 a.m.**

**S4:404—Cell Therapy Counteracts Disease Phenotypes in a Mouse Model of FSHD**

**Karim Azzag**, Darko Bosnakovski, Sudheer Tungtur, Peter Salama, Michael Kyba, Rita Perlingeiro

**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, Karlien 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, Jay Han, Anthony Accorsi, Jordi Diaz, Miriam Freimer, Angela Genge, Summer Gibson, Nuria Gomez, Namita Goyal, Johanna Hamel, Lawrence Hayward, John Jiang, Nicholas Johnson, Joost Kools, David Reyes Leiva, Doris Leung, Hanns Lochmuller, Samantha Lorusso, Michelle L. Mellion, Alan Pestronak, L. Alejandro Rojas, Sabrina Sacconi, Perry Shieh, Jennifer Shoskes, **Jeffrey Statland**, S.H. Subramony, Rabi Tawil, Baziel van Engelen, Juan Vilchez, Kathryn 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**



# POSTER PRESENTATIONS

## 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

**P1.01 Single cell sequencing identifies unique transcriptional responses to plasma membrane injury in FSHD**  
Adam Bittel, Surajit Bhattacharya, **Yi-Wen Chen**

**P1.02 Single-nucleus RNA-Seq reveals cellular heterogeneity in facioscapulohumeral muscular dystrophy in late myogenic stage**  
**Dongxu Zheng**, Anita van den Heuvel, Ahmed Mahfouz, Annelot Wondergem, Susan Kloet, Judit Balog, Baziel van Engelen, Rabi Tawil, Stephen Tapscott, Silvère Van der Maarel

**P1.03 Generation of a craniofacial muscle model of FSHD from iPSCs**  
Dongsheng Guo, Oliver King, **Lawrence Hayward**, Charles P. Emerson Jr.

**P1.04 The inflammatory muscle phenotype of uninduced ACTA1-MCM;FLEXD mice**  
**Jessica de Greef**, Linde Bouwman, Bianca den Hamer, Silvère Van der Maarel

**P1.05 Towards a muscle-targeted delivery of antisense agents against DUX4**  
**Maëlle Limpens**, Aline Derenne, Carmen Burtea, Alexandre Legrand, Anne-Emilie Decleves, Frédérique Coppée, Alexandra Tassin

**P1.06 WITHDRAWN**

**P1.07 p38-independent DUX4 regulatory mechanisms in FSHD myotubes**  
**Rajanikanth Vangipurapu**, Francis M. Sverdrup

**P1.08 SLC34A2 as a protein biomarker of FSHD**  
**Maria Traficante**, Andrea O'Neill, Ujwala Pimparkar, Rabi Tawil, Jeffrey Statland, Robert Bloch

**P1.09 Dynamic proteome profiling of myoblasts from FSHD patients and their unaffected siblings**  
**Jatin Burniston**, Radoš Stefanović, Adam Bittel, Yi-Wen Chen

**P1.10 Promoting clinical trial readiness to Brazil**  
**Fabio Eliezer Figueiredo**, Cristiane A Martins Moreno

**P1.11 Transient DUX4 expression leads to muscle degeneration**  
David Oyler, Ana Mitanoska, Ahmed Shams, Natasha Santos, Natalie Xu, Jasmine Gulik, MacKenzie Molina, Erik Toso, Michael Kyba, **Darko Bosnakovski**

## GENETICS & EPIGENETICS

**P2.12 Generation of human skeletal myocyte models harboring SMCHD1 and/or D4Z4 mutations reveals critical roles of epigenetic modifiers for stable FSHD phenotype**  
**Nam Nguyen**, Xiangduo Kong

**P2.13 Methylation analysis of proximal region on D4Z4 repeats in FSHD1 patients compared to healthy individuals**  
**Ceren Hangul**, Öznur Tokta, Sibel Berker Karauzum, Hilmi Uysal, Filiz Koc

**P2.14 Cis D4Z4 repeat duplications in FSHD**  
**Richard Lemmers**, Patrick van der Vliet, Silvère van der Maarel, Jan de Bleecker, Ludo van der Pol, Corrie van Erasmus, Marc D'Hooghe, Peter van den Bergh, Baziel van Engelen, Jeffrey Statland, Rabi Tawil, Nicol Voermans, Sabrina Sacconi, John Vissing, Silvère M van der Maarel

**P2.15 Maternal SMCHD1/LRIF1 haploinsufficiency triggers homeotic transformations in genetically wild-type offspring**  
**Shifeng Xue**, Frederique Magdinier, Bruno Reversade

## PATHOLOGY & DISEASE MECHANISMS

- P3.16 Generation of mouse artificial chromosome carrying human chromosome 4q35 for a novel FSHD1 mouse model**  
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, AnnaGreco, Leo Joosten, Baziël van Engelen, Peter Zammit
- 
- P3.18 Relationship between DUX4 and Hypoxia-Inducible Factor (HIF1 $\alpha$ ) in human and murine muscle cells in vitro and in vivo**  
Thuy Hang Nguyen, Alexandre Legrand, Anne-Emilie Decleves, Philipp Heher, Alexandra Belayew, Christopher Banerji, Peter Zammit, Alexandra Tassin
- 
- P3.19 Fibro-adipogenic progenitors and the progression of the FSHD myopathy**  
Carlo Serra, Kathryn Wagner, Thomas Lloyd
- 
- P3.20 DUX4, nucleolar stress, and FSHD myopathy**  
Carlo Serra, Kathryn Wagner, Thomas Lloyd
- 
- P3.21 Innate immunity model of FSHD muscle pathology activates complement genes**  
Katelyn Daman, Jing Yan, Oliver King, Michael Brehm, Charles P. Emerson Jr.
- 
- P3.22 Intramuscular fibrosis correlates with disease activity and progression in facioscapulohumeral muscular dystrophy patients**  
Elvira Ragozzino, Sara Bortolani, Lorena Di Pietro, Ornella Parolini, Mauro Monforte, Giorgio Tasca, Enzo Ricci
- 
- P3.23 Non-myogenic mesenchymal cells contribute to muscle degeneration in facioscapulohumeral muscular dystrophy patients**  
Lorena Di Pietro, Flavia Giacalone, Elvira Ragozzino, Valentina Saccone, Marco De Bardi, Mario Picozza, Wanda Lattanzi, Enrico Guadagni, Sara Bortolani, Giorgio Tasca, Enzo Ricci, Ornella Parolini
- 
- P3.24 Interaction between mesenchymal stem cells and myoblasts contributes to the FSHD phenotype**  
Yegor Vassetzky, Ekaterina Kiseleva, Olesya Serbina, Anna Karpukhina
- 

## INTERVENTIONAL STRATEGIES

- P4.25 WITHDRAWN**
- 
- P4.26 Hit-and-run silencing of endogenous DUX4 by targeting DNA hypomethylation on D4Z4 repeats in in vitro FSHD-iPSC model**  
Mitsuru Sasaki-Honda, Junjie He, Hidetoshi Sakurai
- 
- P4.27 Improving FSHD RNAi gene therapy using myotropic MyoAAVs**  
Lindsay Wallace, Tessa Riley, Matthew Guggenbiller, Gholamhossein Amini Chermahini, Scott Harper
- 
- P4.28 Identification and targeting of hypoxia signaling for translational potential in facioscapulohumeral muscular dystrophy**  
Justin Cohen, Vincent Ho, Alec Desimone, Monkol Lek, Angela Lek
- 
- P4.29 An AAV-shRNA DUX4-based therapy to treat facioscapulohumeral muscular dystrophy (FSHD)**  
Virginie Mariot, Eva Sidlauskaitė, Laura Le Gall, Emilio Corbex, Julie Dumonceaux
- 
- P4.30 Development of safe and efficacious RNA therapeutics for FSHD**  
Christian Kinney, Anthony Saleh
-



# POSTER PRESENTATIONS

## CLINICAL STUDIES & OUTCOME MEASURES

**P5.31 Motor outcomes to validate evaluations in facioscapulohumeral muscular dystrophy (MOVE FSHD): protocol for an observational study**

**Michaela Walker**, Russell Butterfield, John Day, Kate Eichinger, Bakri Elsheikh, Anna Faino, Seth Friedman, Kiley Higgs, Nicholas Johnson, Peter Jones, Doris Leung, Leann Lewis, Bill Martens, Dennis Shaw, Perry Shieh, Subramony Subramony, Jaya Trivedi, Leo Wang, Mathew Wicklund, Rabi Tawil, Jeffrey Statland

**P5.32 Muscle imaging in facioscapulohumeral muscular dystrophy (FSHD): relevance for clinical trials. Report from the 265th ENMC Workshop**

**Giorgio Tasca**, Shahram Attarian, John Vissing, Jordi Diaz-Manera, Nicol Voermans

**P5.33 The face of facioscapulohumeral muscular dystrophy: exploring facial muscle involvement using ultrasound**

**Sanne Vincenten**, Karlien Mul, Nicol Voermans, Nens van Alfen, Baziel van Engelen

**P5.34 Analyzing the impact of FSHD on patient outcomes**

**Elan Schonfeld**, Charulatha Nagar

**P5.35 A world-wide survey of standardised outcome measure use in FSHD clinical care**

**Katy de Valle**, Jenny McGinley, Fiona Dobson, Monique Ryan

**P5.36 Longitudinal assessment of facial weakness in facioscapulohumeral muscular dystrophy by physicians and patients**

**Karlien Mul**, Tom Loonen, Sanne Vincenten, Sjan Teeselink, Nicol Voermans, Thomas Maal, Baziel van Engelen

**P5.37 Muscle ultrasound in an open-label study of losmapimod in subjects with FSHD1**

**Joost Kools**, Nicol Voermans, Karlien Mul, John Jiang, Jennifer Shoskes, Kelly Marshall, Michelle L. Mellion, Baziel van Engelen, Markus Karlsson

**P5.38 Feasibility of measuring functional performance of FSHD patients using wearable sensors to quantify physical activity**

**Joost Kools**, Nicol Voermans, Karlien Mul, Michelle L. Mellion, John Jiang, Jennifer Shoskes, Kelly Marshall, David Jackson, Yuxi Zhao, Anil Tarachandani, Joanita Figueredo, Damien Eggenspieler, Baziel van Engelen

**P5.39 Living with FSHD during the pandemic corona outbreak in the Netherlands: pitfalls and challenges of COVID-19 in FSHD**

**Joost Kools**, Johanna Deenen, Anna Greco, Renée Thewissen, Wiecke Van de Put, Anke Lanser, Nicol Voermans, Mara Tihaya, André Verbeek, Silvère Van der Maarel, Leo Joosten, Baziel van Engelen

**P5.40 The UK FSHD Patient Registry: a key tool linking patients with national and international research projects**

**Helen Walker**, Richard Orrell, Chiara Marini-Bettolo, Andrew Graham, Kate Adcock, Suzanne Watt, Peter Lunt, Fiona Norwood, Mark Roberts, Tracey Willis, Emma Matthews, Robert Muni-Lofra

**P5.41 Prevalence and impact on quality of life of gastrointestinal and genitourinary symptoms in facioscapulohumeral muscular dystrophy**

**Michael Cole**, June Kinoshita, Angelena Edwards, Christopher Cooper, Eyad Hanna, M. Bridget Zimmerman, Katherine Mathews

**P5.42 Inpatient admissions and emergency department visits for patients with facioscapulohumeral muscular dystrophy (FSHD): a real-world retrospective data analysis of pre- and post-diagnosis events**

Chamindra Konersman, **Kathryn Munoz**, Richard Brook, Nathan Kleinman, Kelly DiTrapani, Bradley McEvoy, Alissa Peters, Chao-Yin Chen, Teresa Brandt, Mark Stahl



**P5.43 Design of REACH: Phase 3 randomized, double-blind, placebo-controlled, 48-week study of the efficacy and safety of losmapimod in FSHD**

Christopher Morabito, Rabi Tawil, Jay Han, Leo Wang, John Vissing, Baziel van Engelen, Jeffrey Statland, Michelle L. Mellion, **Jennifer Shoskes**, John Jiang, Jennifer Webster

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**P5.44 Annualized rates of change from a Phase 2, randomized, double-blind, placebo-controlled, 48-week study of losmapimod in subjects with FSHD: ReDUX4**

Christopher Morabito, Sabrina Sacconi, Jordi Diaz, David Reyes Leiva, Doris Leung, Kathryn Wagner, Angela Genge, Juan Vilchez, Nuria Gomez, Miriam Freimer, Samantha Lorusso, Hanns Lochmuller, Joost Kools, Baziel van Engelen, Namita Goyal, Perry Shieh, S.H. Subramony, Jeffrey Statland, Lawrence Hayward, Johanna Hamel, Rabi Tawil, Summer Gibson, Leo Wang, Nicholas Johnson, Alan Pestronak, Michelle L. Mellion, Anthony Accorsi, **Jennifer Shoskes**, John Jiang, L. Alejandro Rojas

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**P5.45 Understanding falls in FSHD**

**Enrico Bugiardini**, Kate Eichinger, Michael Hanna, Gita Ramdharry, Michael McDermott, Kiley Higgs, Michaela Walker, Leann Lewis, Bill Martens, Doris Leung, Sabrina Sacconi, Karlien Mul, Valeri Sansone, Leo Wang, Perry Shieh, Bakri Elsheikh, Russell Butterfield, Nicholas Johnson, Rabi Tawil, Jeffrey Statland, the ReSolve Investigators of the FSHD CTRN

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**P5.46 TREAT-NMD FSHD Global Registry Network: a collaboration of neuromuscular and FSHD patient registries**

**Ben Porter**, Neil Bennett, David Allison, Craig Campbell, Michela Guglieri, Anna Ambrosini, Rossella Tupler

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**P5.47 A case story: supervised FSHD patient self-analysis of its respiratory data**

**Patrick Valentin**, Frederic Lofaso

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**P5.48 Longitudinal whole-body MRI and muscle function in FSHD1**

**Doris Leung**, Sharanya Suresh, Shivani Ahlawat, Alex Bocchieri, Vishwa Parekh, Vladimir Braverman, Michael Jacobs

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**P5.49 The FSHD Composite Outcome Measure (FSHD-COM) is reliable, valid, and measures disease progression**

**Katy Eichinger**, Michael McDermott, Kiley Higgs, Michaela Walker, Leann Lewis, Bill Martens, Doris Leung, Nikia Stinson, Megan McNerney, Sabrina Sacconi, Jeremy Garcia, Victor De Paz Benito, Karlien Mul, Valeria Sansone, Elena Carraro, Stefano Becchiati, Maria Frisoni, Leo Wang, Catherine Kieu, Perry Shieh, Christ Skura, Bakri Elsheikh, Kristina Kelly, Andrea Jaworek, Samantha LoRusso, Russell Butterfield, Amelia Wilson, Melissa McIntyre, Nicholas Johnson, Amanda Butler, Aileen Jones, Melissa Hayes, Sandhya Sasidharan, Lindsay Baker, Rabi Tawil, Jeffrey Statland, the ReSolve Investigators of the FSHD CTRN

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**P5.50 Understanding the perseverance of the muscular dystrophy community one year into the COVID-19 pandemic**

**Leann Lewis**, Katy Eichinger, Nuran Dilek, Kiley Higgs, Michaela Walker, David Palmer, John Cooley, Nicholas Johnson, Rabi Tawil, Jeffrey Statland

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**P5.51 Clinical and molecular evaluation of FSHD patients in Turkey**

**Serpil Eraslan**, Sahin Avci, Ilker Eren, Gulshan Yunisova, Piraye Oflazer, Mehmet Demirhan, Hulya Kayserili

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**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

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**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

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# COMPLEX RESORT MAP



## RESTAURANTS & LOUNGES

- 1 Zeta Asia
- 2 MUSE
- 3 Harvest Bistro
- 4 La Luce®
- 5 Beech®
- 6 MYTH Bar
- 7 Aquamarine
- 8 Bull & Bear®
- 9 Peacock Alley®
- 10 Oscar's®
- 11 Sir Harry's Lounge
- 12 The Clubhouse Grille

## FUNCTION SPACE

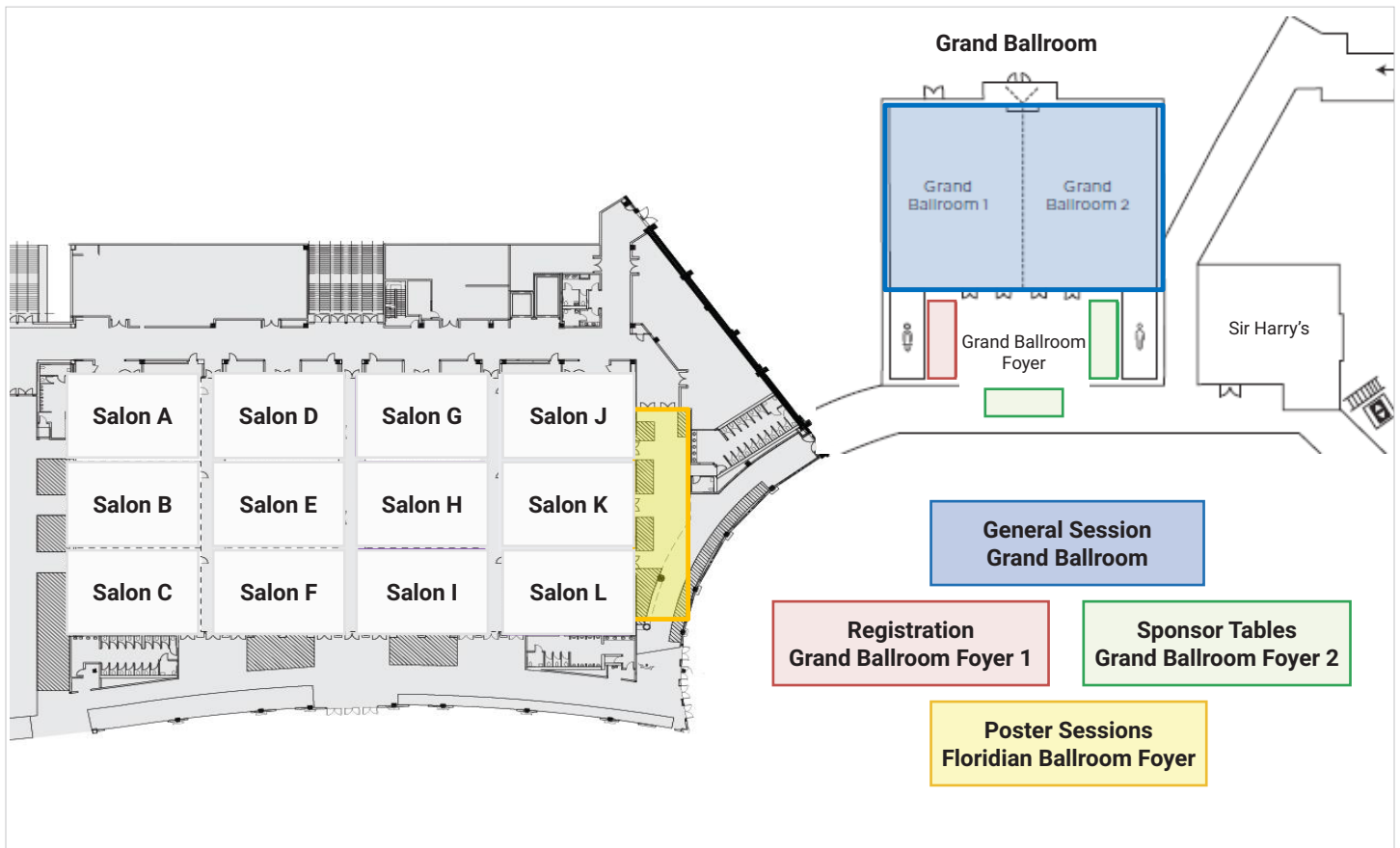
- 13 Waldorf Astoria Meeting Rooms
- 14 Waldorf Astoria Grand Ballroom
- 15 Promenade
- 16 Signature Island
- 17 Signia by Hilton Meeting Rooms
- 18 Floridian Ballroom
- 19 Bonnet Creek Ballroom

## AMENITIES & TRANSPORTATION

- 20 Signia by Hilton Pool & Lazy River
- 21 Signia by Hilton Water Slide
- 22 Signia by Hilton Private Cabanas
- 23 Waldorf Astoria Private Cabanas
- 24 Waldorf Astoria Pool Cabanas
- 25 Waldorf Fitness Center
- 26 Signia by Hilton Fitness Center
- 27 Waldorf Astoria Spa
- 28 Full-Service Business Center
- 29 Self-Service Business Center (24-Hour)
- 30 Waldorf Astoria® Golf Pro Shop
- 31 Waldorf Astoria® Golf Club
- 32 FunZone Arcade
- 33 Disney Store
- 34 Sundries Shop
- 35 Daniella Ortiz
- 36 Golf Pavilion
- 37 Golf Pavilion Gardens
-  Shuttle Pick-Up Station
-  Signia by Hilton & Conference Parking



# ROOM LAYOUT



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