



# PREFACE

June 8–9, 2018 | Las Vegas, Nevada

DEAR COLLEAGUES,

Welcome to the FSH Society 2018 International Research Congress & Research Planning Meetings!

This workshop brings together clinicians, scientists, industry and patient representatives, and policy makers to discuss the latest developments in facioscapulohumeral muscular dystrophy (FSHD). For more than two decades, this gathering has provided the FSHD community with a forum to present and discuss new findings, reinforce collaborative efforts, facilitate new initiatives, and coordinate research, therapeutic, and clinical activities.

Exceptional scientific progress has been made in recent years and months in our understanding of the disease. This is a critically important time for the community to convene and discuss new data and advances in FSHD; develop strategies to verify and independently corroborate the findings; discuss focusing efforts and resources in the preclinical gap and translational phase of research; improve diagnostic techniques and criteria for FSHD; and consider and evaluate with industry how to move forward with accelerating new and existing therapies.

Over the two days, we will revisit the priority areas identified at the most recent 2016 meeting and discuss what we have achieved, evaluate the gaps that need addressing, and decide where we need to focus and invest intellectual, scientific, and financial resources. By the end of day two we should be able to identify whether any of the previous year's priority areas should change or be modified, and outline a new list/set of priority areas to be considered.

This meeting is organized by the FSH Society and sponsored by Acceleron, Association Française Contre les Myopathies Muscular Dystrophy Association, FSHD Canada Foundation, Fulcrum Therapeutics, Genea Biocells, Genomic Vision, Muscular Dystrophy Association, NIH NICHD UMass Senator Paul Wellstone MD Cooperative Research for FSHD, Sanofi US Services Inc., Ultragenyx, and University of Nevada, Reno. We thank our sponsors for their generous financial support.

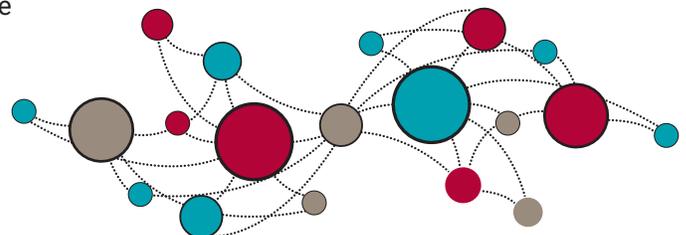
Friday platform and poster presentations are considered confidential scientific presentations that contain unpublished data and should not be photographed or incorporated in newsletters or used in any other manner without the permission of the reporting scientists. There is an assumption of confidentiality for Saturday's discussions as in other scientific meetings. Saturday's discussion will be recorded as in past years to help generate a full set meeting transcripts.

It is truly a pleasure to come together to accelerate solutions for FSHD. Thank you for your extraordinary efforts and hard work on behalf of patients and their families.

Sincerely,

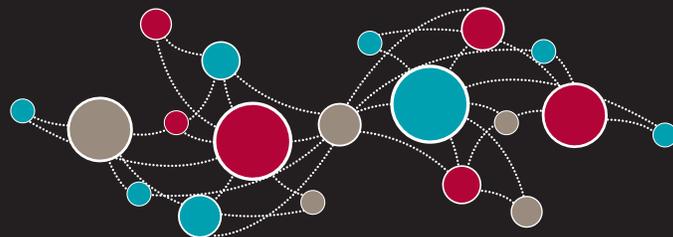
Members of the 2018 IRC Meeting Organizing Committee

Daniel Paul Perez, *FSH Society*,



**Wifi network:** FSH Society  
**Password:** curefshd

# SCHEDULE



## Friday, June 8, 2018—El Dorado Ballroom

### BREAKFAST

**7:00–7:55 a.m.**

El Dorado Ballroom Foyer & Carson City Ballroom

### WELCOME

**7:55–8:00 a.m.**

#### Opening remarks

James Chin, Mark Stone, *2018 IRC Meeting Organizing Committee*

### REVIEW OF 2017

**8:00–8:10 a.m.**

**Review 2017/2018 priorities stated by FSHD workshop in 2016**

Moderators: Michael Altherr, Stephen Tapscott, others TBD

### PLATFORM SESSION 1—8:10–9:25 a.m.

**Genetics, epigenetics, and related syndromes and diseases, cancers, and BOSMA Arhinia**

Moderators: Silvère van der Maarel and Marnie Blewitt

**8:10–8:25 a.m.**

Brand (presenter)/Talkowski: **Lessons in oligogenetics and pleiotropy: identical SMCHD1 alleles can be associated with arhinia, Bosma syndrome, FSHD2, comorbidities, or no phenotype at all**

**8:25–8:40 a.m.**

Mohassel/Shaw (presenter): **Deep neuromuscular phenotyping of arhinia patients with SMCHD1 mutations reveals a mild myopathy distinct from FSHD2**

**8:40–8:55 a.m.**

Jansz/Blewitt (presenter): **The epigenetic repressor, FSHD2 gene and FSHD1 modifier SMCHD1 functions by mediating long range chromatin interactions**

**8:55–9:05 a.m.**

Nguyen/Magdinier (presenter): **Genetic variability and identification of complex genotypes in FSHD patients by molecular combing**

**9:05–9:15 a.m.**

Lemmers (presenter)/van der Maarel: **Cis D4Z4 repeat duplications associated with FSHD2**

**9:15–9:25 a.m.**

Campbell (presenter)/Tapscott: **Identifying mechanisms that regulate DUX4 and the D4Z4 macrosatellite repeats**

**9:25–9:35 a.m.**

Discussion

### PLATFORM SESSION 2— 9:35–11:00 a.m.

**The role of DUX4 in development and disease**

Moderators: Peter Zammit and Stephen Tapscott

**9:35–9:45 a.m.**

de Morrée (presenter)/Rando: **U1 snRNA controls alternative polyadenylation of Pax3 in muscle stem cells**

**9:45–10:00 a.m.**

Banerji (presenter)/Zammit: **Dynamic transcriptomic and morphological analysis of FSHD atrophic myogenesis reveals a correctable defect in mitochondrial biogenesis**

**10:00–10:15 a.m.**

Kyba (presenter)/Aihara: **Structural and functional studies on DUX4 in human myogenesis**

**10:15–10:30 a.m.**

Eidahl (presenter)/Harper: **Regulation of facioscapulohumeral muscular dystrophy candidate protein DUX4**

**10:30–10:40 a.m.**

DeSimone (presenter)/Emerson: **Identification of a DUX4-interacting protein and the hyaluronic acid pathway as novel therapeutic targets for FSHD**

**10:40–10:50 a.m.**

Mariot/Dumonceaux (presenter): **Myostatin expression in neuromuscular diseases: challenges and hopes**

**10:50–11:00 a.m.**

Saad (presenter)/Harper: **The natural microRNA miR-675 reduces DUX4 expression and toxicity in vitro**

**11:00–11:15 a.m.**

Discussion

# SCHEDULE

Friday, June 8, 2018—El Dorado Ballroom

*continued*

## PLATFORM SESSION 3—11:15–12:20 p.m.

**Preclinical studies in FSHD, including cellular (myoblasts/iPS) and animal models**

Moderators: Peter Jones and Michael Kyba

**11:15–11:30 a.m.**

Chen/Hayward (presenter): **Single-cell transcriptome heterogeneity in myogenic cells from individuals with FSHD**

**11:30–11:45 a.m.**

van den Heuvel (presenter)/van der Maarel: **Single-cell RNA-sequencing in facioscapulohumeral muscular dystrophy disease etiology and development**

**11:45 a.m.–12:00 p.m.**

Giesige (presenter)/Harper: **AAV.RNAi and follistatin gene therapy development in the TIC-DUX4 Mouse Model of FSHD**

**12:00–12:10 p.m.**

Daman (presenter)/Emerson: **An FSHD cell xenograft assay for drug development**

**12:10–12:20 p.m.**

Chen (presenter)/Yokota: **Systemic delivery of LNA gapmers targeting DUX4 improved muscle function in FLExDUX4 mice**

**12:20–12:30 p.m.**

Discussion

## LUNCH AND POSTER VIEWING

**12:30–2:00 p.m.**

Lunch in Carson City Room;  
posters in El Dorado Ballroom Foyer

## PLATFORM SESSION 4—2:00–3:00 p.m.

**Clinical studies and clinical trial**

Moderators: Jeffrey Statland and Baziel van Engelen

**2:00–2:15 p.m.**

Horlings (presenter)/van Engelen: **Clinical outcome measures, muscle imaging and (epi)genetic testing in a large cohort of FSHD patients**

**2:15–2:30 p.m.**

Sacconi (presenter)/van der Maarel: **FSHD1 and FSHD2 form a disease continuum**

**2:30–2:40 p.m.**

Hamel (presenter)/Statland: **MRI correlates to electrical impedance myography in facioscapulohumeral muscular dystrophy**

**2:40–2:50 p.m.**

Zhang (presenter)/Zhang: **Accurate molecular diagnosis of facioscapulohumeral muscular dystrophy in a cohort of 37 Chinese patients**

**2:50–3:00 p.m.**

Zheng (presenter)/Kong: **A case of first trimester prenatal diagnosis for FSHD1 using Karyomapping and single-molecule optical mapping**

**3:00–3:15 p.m.**

Discussion

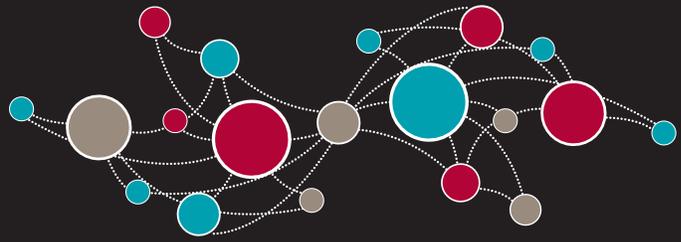
## PLATFORM SESSION 5—3:15–4:20 p.m.

**Industry aspects and therapy development (screens)**

Moderators: Peter Jones and George Padberg

**3:15–3:25 p.m.**

Statland (presenter)/Attie: **Preliminary results from a dose-escalation phase 2 study to evaluate ACE-083, a local muscle therapeutic, in patients with facioscapulohumeral muscular dystrophy**



**3:25–3:40 p.m.**

Rojas (presenter)/Cacace: **Pharmacological inhibition of DUX4 expression rescues FSHD pathophysiology in FSHD skeletal muscle myotubes**

**3:40–3:50 p.m.**

Hupper/Clarke (presenter): **A low molecular weight compound screen in FSHD patient myotubes identifies modulators of Dux4 activity and novel mechanisms of action**

**3:50–4:00 p.m.**

Cruz/Clarke (presenter): **Protein kinase A activation inhibits DUX4 gene expression in myotubes from patients with FSHD**

**4:00–4:10 p.m.**

Lu-Nguyen (presenter)/Poplewell: **In vivo assessment of antisense therapy for Facioscapulohumeral muscular dystrophy**

**4:10–4:20 p.m.**

Rickard/Schmidt (presenter): GBC0905: **A novel targeted therapeutic agent to treat facioscapulohumeral muscular dystrophy**

**4:20–4:30 p.m.**

**Discussion**

**ASSEMBLY SESSION**

**4:20–4:30 p.m.**

**Discussion and review of Saturday's (day 2) agenda**  
Moderators: 2018 IRC Meeting Organizing Committee

**ADJOURN**

**5:00 p.m.**

**Saturday, June 9, 2018—El Dorado Ballroom**

**REGISTRATION & BREAKFAST**

**7:30–8:30 a.m.**

El Dorado Ballroom Foyer & Carson City Ballroom

**PLANNING AND PROBLEM-SOLVING SESSION**

Moderated discussion sessions with entire group of attendees based on data presented at day 1. Co-chairs and organizers will meet end of day Friday and/or Saturday morning before the session to help identify specific topics of interest to lead the discussion. The goals are to 1) help identify and troubleshoot bottlenecks; and 2) define the research/clinical priorities for the next year 2018–2019.

**WELCOME**

**8:30–8:35 a.m.**

2018 IRC Meeting Organizing Committee

**PLANNING AND PROBLEM-SOLVING SESSION(S)**

**8:35–11:15 a.m.**

**International “lab meeting.” Discussion to identify and troubleshoot bottlenecks, and define the research/clinical priorities going forward**

Moderators: Michael Altherr, 2018 IRC Meeting Organizing Committee, and others TBD

**11:15–11:45 a.m.**

**Finalizing listing of items, areas, and priorities**

**LUNCH**

**11:45 a.m.–12:45 p.m.**

Lunch located in Carson City Ballroom; posters should be taken down.

**ADJOURN**

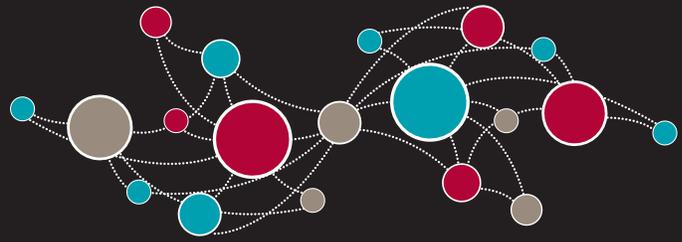
**12:45 p.m.**

# POSTER SESSIONS

## POSTER NUMBER

## AUTHORS AND TITLES

- 30** **Calandra/Deidda (presenter)**  
Large-scale methylation analysis in facioscapulohumeral muscular dystrophy (FSHD)
- 31** **Cammish/Orrell (presenter)**  
The UK FSHD Patient Registry: a key tool in the facilitation of clinical research
- 32** **Chang**  
Testing the potential for comorbidity of FSHD with arhinia using inducibility of DUX4 expression in dermal fibroblasts
- 33** **Choi**  
Establishment of FSHD-PAX7 genetic reporter lines to study function of muscle stem cells in FSHD
- 34** **Choi/Lim (presenter)**  
Modular platform for the myogenesis of human embryonic stem cells by using multiple genetic reporter lines
- 35** **Ciskewski/Popplewell (presenter)**  
Novel epigenetic mechanisms regulating DUX4 expression
- 36** **Claus**  
Direct interaction of DUX4/4c with the multifunctional protein C1QBP
- 37** **Coulis**  
Overexpression of DUX-4 induces muscle Tregs: a potential role for the immune system in FSHD
- 38** **Denny**  
High-density lipoproteins protect against DUX4-mediated damage in a lentiviral model of FSHD
- 39** **Dion/Robin (presenter)**  
Implication of SMCHD1 in D4Z4 epigenetic dynamics: lesson from iPSCs
- 40** **Han**  
Longitudinal study of Kinect-based upper extremity reachable workspace in FSHD
- 41** **Hiramuki**  
A mapping study of SMCHD1 identifies the region of nuclear localization, dimerization, and protein cleavage
- 42** **Homma**  
DUX4 alters mRNA splicing of TDP-43 target
- 43** **Jones, T**  
The FLExDUX4 transgenic mouse can be used to develop FSHD-like mouse models with pathophysiology ranging in severity
- 44** **Kazakov**  
Some problems connected with AD FSHD classification
- 45** **Lopez**  
Autologous stem cell treatment in FSHD. preliminary report
- 46** **Lunt**  
No evidence for altered incidence of cancer in FSHD



## POSTER NUMBER

## AUTHORS AND TITLES

- | POSTER NUMBER | AUTHORS AND TITLES   |
|---------------|--|
| 47            | <b>Maruyama</b><br>Development of LNA and 2'-MOE gapmers to treat facioscapulohumeral muscular dystrophy   |
| 48            | <b>Mueller</b><br>Xenografting human muscle stem cells into mice to study FSHD   |
| 49            | <b>Pakula</b><br>The role of estrogen regulation in FSHD-1   |
| 50            | <b>Rashnonejad</b><br>AAV.U7-sRNA-mediated exon skipping of the toxic DUX4 gene as a promising therapeutic approach for facioscapulohumeral muscular dystrophy |
| 51            | <b>Robertson</b><br>Measurement of evidence of DUX4 as a proof of concept biomarker for FSHD clinical trials   |
| 52            | <b>Sanson/Sacconi (presenter)</b><br>Self-report questionnaire vs. clinical evaluation form in the French National FSHD Registry: a statistical comparison     |
| 53            | <b>Teveroni/Moretti (presenter)</b><br>Set-up of an in vivo model of facioscapulohumeral muscular dystrophy (FSHD) based on human perivascular cells           |
| 54            | <b>van der Stoep</b><br>Evaluation of FSHD1 testing in diagnostics using FiberVision molecular combing technology  |

## FSH SOCIETY 2018 INTERNATIONAL RESEARCH CONGRESS & RESEARCH PLANNING MEETINGS

### Co-chairs:

Marnie Blewitt, PhD, *Walter + Eliza Hall Institute of Medical Research*  
Peter L. Jones, PhD, *University of Nevada, Reno*  
Michael Kyba, PhD, *Lillehei Heart Institute, University of Minnesota*  
Jeffrey Statland, MD, *University of Kansas*  
Stephen J. Tapscott, MD, PhD, *Fred Hutchinson Cancer Research Center*  
Silvère van der Maarel, PhD, *Leiden University Medical Center*  
Baziel van Engelen, MD, PhD, *Radboud University Nijmegen Medical Centre*  
Peter Zammit, PhD, *King's College London*

### Organizers:

Daniel Paul Perez & FSH Society Scientific Advisory Board

The FSH Society would like to thank our Gold Sponsors:



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The FSH Society is an independent, non-profit 501(c)(3) and tax-exempt U.S. corporation organized to address issues and needs specifically related to facioscapulohumeral muscular dystrophy (FSHD). Contributions and financial donations are acknowledged for tax purposes. All inquiries should be addressed to: FSH Society, Daniel Paul Perez, 450 Bedford Street, Lexington, MA 02420. Phones: (781) 301-6650 and (781) 275-7781, fax: (781) 862-1116, email: [daniel.perez@fshsociety.org](mailto:daniel.perez@fshsociety.org), website: <http://www.fshsociety.org>