

26th Annual
**FSHD International
Research Congress**
PROGRAM



JUNE 19-20, 2019 | PALAIS DU PHARO | MARSEILLE, FRANCE



BIENVENUE!

The FSH Society's annual research conference is the premier global platform for the discussion and dissemination of state-of-the-art research on facioscapulohumeral muscular dystrophy (FSHD). Researchers who have networked at this yearly gathering since 1994 have helped to drive fundamental discoveries, with a broad consensus on a "central hypothesis" of the genetic basis of FSHD, development of cellular and animal models, identification of therapeutic targets, and a flowering of ideas for treating FSHD. We are nearing the completion of a Phase 2 clinical trial of a myostatin inhibitor, and the first disease-modifying therapy for FSHD will be launching a Phase 2 trial this year.

The FSH Society's annual research conference is being held for the first time in Europe, to reflect the tremendous contributions of European centers of excellence in FSHD. To keep pace with the expansion of FSHD research, this meeting has grown, too, from one day to two full days. The program committee has particularly reached out to attract clinical researchers, as advancement of drug development brings greater urgency to improving our understanding of the clinical features of FSHD, genotype-phenotype correlations, natural history, and evaluation of disease progression.

With the recent advances in FSHD studies now including large sets of data, greater availability of biomaterials from multiple large research initiatives, and the rapid approach of more clinical trials, the FSHD International Research Congress serves a more important role than ever: to ensure dissemination of the latest ideas and discussion of the field's needs and priorities, combined with collaboration and coordination, to speed up progress toward delivering effective treatments to our patients and families.

We sincerely thank our local host institutions: Marseille Medical Genetics; INSERM; GIPTIS (Genetics Institute for Patients, Therapies, Innovation and Science); the Department of Medical Genetics and Centre de Référence des maladies neuromusculaires et de la SLA, Hôpital de la Timone, Marseille.

We thank you for participating to make such an exciting and dynamic program, and hope that you enjoy the 2019 IRC as much as we have enjoyed planning it.

Sincerely,

THE 2019 IRC PROGRAM COMMITTEE

June Kinoshita
ORGANIZATIONAL CHAIR

Frédérique Magdinier
CO-CHAIR

George Padberg
CO-CHAIR

Alexandra Belayew

Sabrina Sacconi

Stephen Tapscott

Rabi Tawil

Rossella Tupler

Peter Zammit



MICHEL FARDEAU

Professor Fardeau is emeritus research director at the Centre national de la recherche scientifique (CNRS) and honorary professor at the CNAM. In 1977, as a young research director, Prof. Fardeau became head of the biology and neuromuscular pathology unit at CNRS, which he directed until 1996. Initially a small team, four people established the first unit working specifically in the field of muscle diseases: the electron microscopy department at La Salpêtrière. The team grew gradually, to the point that it moved from La Salpêtrière to Fer à Moulin, and then it became necessary to build the Institute of Myology, with a staff that now exceeds several hundred members, which Prof. Fardeau served as medical and scientific director until his retirement in 2006. Prof. Fardeau has received numerous awards and international recognition for his work. In 2015, Prof. Fardeau received the Grand Medal of the French National Academy of Medicine. He was honored for his outstanding career that has been entirely devoted to the biology and pathology of muscle. He is currently a member of the scientific council of the Association Française contre les Myopathies.



BRADLEY R. CAIRNS

Dr. Cairns received his B.S. (Honors) in chemistry from Lewis and Clark College in 1987. He received his PhD in cell biology in 1996 from Stanford University, where he worked with Nobel Laureate Roger Kornberg on signal transduction and chromatin remodeling. Dr. Cairns received formal postdoctoral training with Fred Winston, PhD, in the Department of Genetics at Harvard Medical School (funding from the Leukemia Society of America), where Dr. Cairns continued to study chromatin remodeling complexes. In 1998, he joined the faculty of the Department of Oncological Sciences at the University of Utah School of Medicine. In 2000, he was appointed as an investigator with the Howard Hughes Medical Institute. He is currently professor and chair of the Department of Oncological Sciences, and is the Jon and Karen Huntsman Presidential Professor in Cancer Research and senior director of basic science at the Huntsman Cancer Institute. He is co-leader of the Nuclear Control of Cell Growth and Differentiation Program. He was elected to the American Academy of Arts and Sciences in 2017. The Cairns lab strives to understand chromatin-transcription relationships – with an emphasis on development and cancer – and effectively utilizes biochemistry, genetics, and genomics in multiple model systems.





SCHEDULE DAY 1

PALAIS DU PHARO, SALLE DE LA MAJOR

7:30–8:30

REGISTRATION

Please take breakfast at your hotel before coming to the Palais du Pharo.

8:30–8:35

WELCOME

Mark Stone, president & CEO, FSH Society
George Padberg & Frédérique Magdinier, co-chairs

8:35–9:00

PLENARY: FSHD, the patients' perspective

9:00–9:20

KEYNOTE: A historical perspective on FSHD

Michel Fardeau, Institute of Myology

9:20–9:30

KEYNOTE: FSHD genetics today

Nicholas Levy, Medical Genetics, Aix-Marseille Université

9:30–9:45

Report on the March 2019 Industry Collaborative Workshop on FSHD Therapeutics

Jamshid Arjomand, chief science officer, FSH Society

9:45–10:10

COFFEE BREAK

Break sponsor: Genomic Vision

10:10–11:10

PLATFORM SESSION 1

The FSHD clinical phenotype

Chair, George Padberg; co-moderator, Peter Lunt

10:10–10:25

1.1 Clinical characteristics of childhood FSHD; implications for trial-readiness.

Nicol Voermans (presenter), RJM Goselink

10:25–10:40

1.2 Clinical examination of scapula function in patients with FSHD.

Jos IJspeert

10:40–10:55

1.3 Facioscapulohumeral muscular dystrophy (FSHD) has a talk with endocrinologic parameters: estradiol, progesterone and testosterone.

Ceren Hangül

10:55–11:10

1.4 High frequency of keratinocyte-related skin diseases in FSHD.

Sabrina Sacconi (presenter), Luisa Villa

11:10–12:10

PLATFORM SESSION 2

Genetics and molecular findings for genotype-phenotype correlations and genetic diagnostics

Chair, Rossella Tupler; co-moderator, Meena Upadhyaya

11:10–11:25

2.1 Large scale genotype-phenotype correlation study in 1,703 carriers of D4Z4 reduced alleles from the Italian National Register for FSHD.

Fabiano Mele

11:25–11:40

2.2 Exploring the pathogenicity of DUX4 permissive and non-permissive 4qA haplotypes in FSHD.

Muriel Kuipers

11:40–11:55

2.3 SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localization of variants in the ATPase domain.

Richard Lemmers

11:55–12:10

2.4 Trans-generational effects in FSHD: clinical and lab evidence for imprinting, possibly cumulative?

Peter Lunt

12:10–13:15

LUNCH: PHAR CLUB

Lunch sponsor: Muscular Dystrophy Association

13:15–15:15

PLATFORM SESSION 3

Molecular mechanisms: DUX4, downstream targets, other players

Chair, Pete Zammit; co-moderator, Michael Kyba

13:15–13:30

3.1 Consequences of DUX4 expression in vitro and in vivo.

Stephen Tapscott (presenter), Rebecca Resnick



SCHEDULE DAY 1

PALAIS DU PHARO, SALLE DE LA MAJOR

13:30–13:45

3.2 Single-cell transcriptomes of myogenic cells in facioscapulohumeral muscular dystrophy.
Laurence Hayward (presenter), Dongsheng Guo

13:45–14:00

3.3 Generation of an iPSC model of FSHD and unveiled aspects of DUX4 expression under genotoxic stresses.
Mitsuru Sasaki-Honda

14:00–14:15

3.4 In vitro challenging of facioscapulohumeral muscular dystrophy macrophages derived monocytes: the role of trained innate immunity in FSHD.
Anna Greco

14:15–14:30

3.5 The interplay between myogenesis and inflammation in FSHD.
Maryna Panamarova

14:30–14:45

3.6 Identification of the hyaluronic acid pathway as a novel therapeutic target for facioscapulohumeral muscular dystrophy.
Alec M DeSimone

14:45–15:00

3.7 The role of mitochondrial reactive oxygen species in pathology of FSHD myogenesis.
Anna Karpukhina

15:00–15:15

3.8 Identification of the DUX4-targeted miRNome from a library of 1,881 natural human miRNAs.
Nizar Y Saad

15:15–15:30

COFFEE BREAK

Break sponsor: University of Nevada, Reno

15:30–17:00

PLATFORM SESSION 4

Molecular mechanisms and animal models

Chair, *Alexandra Belayew*; co-moderator, *Scott Harper*

15:30–15:43

4.1 Transgenic mice expressing tunable levels of DUX4 develop characteristic facioscapulohumeral muscular dystrophy-like pathophysiology ranging in severity.
Peter Jones (presenter), Takako Jones

15:43–15:56

4.2 Muscle xenografts reproduce key molecular features of FSHD.
Robert Bloch

15:56–16:09

4.3 Clinically advanced p38 inhibitors suppress DUX4 expression in cellular and animal models of FSHD.
Fran Sverdrup (presenter), Jonathan Oliva

16:09–16:22

4.4 Examining the etiology of myopathy mediated by the transcription factor Dux.
Paul Gregorevic (presenter), Kevin I Watt

16:22–16:35

4.5 Cellular pathway disruptions in mouse muscle expressing low levels of DUX4 protein.
Joel R Chamberlain (presenter), Maya Zavaljevski

16:35–16:48

4.6 Study of regenerative potential of human perivascular cells expressing DUX4.
Fabiola Moretti (presenter), Silvia Maiullari

16:48–17:00

4.7 Retrotransposon-mediated repression of Dux in early mouse development.
Michelle Percharde

17:00–18:00

BREAK

18:00–20:00

POSTER SESSION

Grand Large (1st level)

19:00–20:00

COCKTAILS

Eugenie Ballroom (ground level)

20:00–22:30

CONFERENCE BANQUET

Eugenie Ballroom, Palais du Pharo



SCHEDULE DAY 2

PALAIS DU PHARO, SALLE DE LA MAJOR

8:25–8:30

WELCOME

Please take breakfast at your hotel before coming to the Palais du Pharo.

George Padberg & Frédérique Magdinier, co-chairs

8:30–9:15

KEYNOTE: The role of DUX4 in development

Brad Cairns, University of Utah

9:15–10:15

PLATFORM SESSION 5

DNA methylation and epigenetics

Chair Frédérique Magdinier; co-moderator, Marnie Blewitt

9:15–9:27

5.1 The crystal structure of SMCHD1's hinge domain and the pathway for SMCHD1 chromatin association reveal a model for how SMCHD1 is targeted to chromatin.

Marnie Blewitt/Kelan Chen

9:27–9:39

5.2 Methylation of the region distal to the D4Z4 array is lower than predicted in FSHD1.

Giancarlo Deidda (presenter), Patrizia Calandra

9:39–9:51

5.3 Identification of a new epigenetic factor required for the aberrant expression of DUX4 in FSHD muscular dystrophy.

Davide Gabellini (presenter), Roberto Giambruno

9:51–10:03

5.4 The D4Z4 macrosatellite sequence as a prototype element for formation of long distance loops: implication in pathologies.

Jerome D Robin (presenter), Marie-Cécile Gaillard

10:03–10:15

5.5 Apabetalone, a CVD phase 3 clinical-stage BET inhibitor, opposes DUX4 expression in primary human FSHD muscle cells.

Christopher D Sarsons

10:15–10:30

COFFEE BREAK

Break sponsor: Filnemus

10:30–11:30

PLATFORM SESSION 6

Muscle pathology and imaging

Chair, Rabi Tawil; co-moderator, Giorgio Tasca

10:30–10:45

6.1 Membrane repair deficits in facioscapulohumeral muscular dystrophy.

Yi-wen Chen (presenter), Sreetama Sen Chandra

10:45–11:00

6.2 Muscle ultrasound is a responsive biomarker in FSHD.

Baziel van Engelen (presenter), Rianne JM Goselink

11:00–11:15

6.3 Long-term follow-up of MRI changes in thigh muscles of patients with facioscapulohumeral dystrophy: a quantitative study.

Emmanuelle Salort-Campana

11:15–11:30

6.4 PATCHS MRI score correlates with clinical severity in facioscapulohumeral muscular dystrophy.

Wenhua Zhu (presenter), Yiqi Liu

11:30–12:15

PLATFORM SESSION 7

Natural history

Chair, Sabrina Sacconi; Co-moderator, Karlien Mul

11:30–11:45

7.1 Clinical categories to describe the phenotypic complexity associated with D4Z4 reduced allele.

Giulia Ricci

11:45–12:00

7.2 Differentiating phenotypes in carriers of 7–8 D4Z4 reduced alleles: experience of the Italian National Registry for FSHD.

Lucia Ruggiero

12:00–12:15

7.3 Longitudinal MRI evaluation of muscle involvement in FSHD.

Mauro Monforte



SCHEDULE DAY 2

PALAIS DU PHARO, SALLE DE LA MAJOR

12:15–12:45

PLATFORM SESSION 8

Registries

Chair, Sabrina Sacconi

12:15–12:45

8.1 Report on national FSHD registries.

Karlien Mul

12:45–14:30

LUNCH BREAK & POSTER SESSION: PHAR CLUB

Lunch sponsor: National Institutes of Health

14:30–16:00

PLATFORM SESSION 9

Therapeutic interventions

Chair, Stephen J. Tapscott; co-moderator, Fran Sverdrup

14:30–14:45

9.1 Dose escalation results from a phase 2 study of ACE-083, a local muscle therapeutic, in patients with facioscapulohumeral muscular dystrophy (FSHD).

Jeffrey M Statland

14:45–15:00

9.2 The discovery of a drug target and development candidate that inhibits the expression of DUX4, the root cause of FSHD.

Owen B Wallace

15:00–15:15

9.3 Translating DUX4-targeted RNAi-based gene therapy for FSHD.

Lindsay Wallace

15:15–15:30

9.4 Inhibition of DUX4 expression with antisense gapmers as a therapy for facioscapulohumeral muscular dystrophy.

Rika Maruyama

15:30–15:45

9.5 Targeting the DUX4 transcriptional mechanism of action.

Michael Kyba (presenter), Darko Bosnakovski

15:45–16:00

9.6 Discovery of novel small molecule treatment options for FSHD.

Joris De Maeyer (presenter), Geese Marcus

16:00–16:15

COFFEE BREAK

Break sponsor: Muscular Dystrophy UK

16:15–17:15

PLATFORM SESSION 10

Clinical evaluation, outcome measures, clinical trial readiness (4 talks, 60 minutes)

Chair, Jeffrey Statland; co-moderator, Baziel van Engelen

16:15–16:30

10.1 Conceptual framework for measurement of treatment effect on DUX4 in losmapimod phase 2 trials.

Lucienne Ronco

16:30–16:45

10.2 Elucidating the extent and pattern of longitudinal upper extremity reachability decline in FSHD, using Kinect sensor-based reachable workspace outcome measure.

Jay Han

16:45–17:00

10.3 Development of an optimized Timed-Up-and-Go Test for the FSHD population.

Maya N Hatch

17:00–17:15

10.4 The facioscapulohumeral muscular dystrophy specific Rasch-built Overall Disability Scale (FSHD-RODS).

Karlien Mul

17:15–18:00

MEETING CONCLUSION & PRIORITIES DISCUSSION

18:00

ADJOURN



POSTER SESSIONS

PALACE DU PHARO, GRAND LARGE (1st level)

Odd-numbered posters on July 19, 18:00-20:00

Even-numbered posters on July 20, 12:45-14:30

POSTER NUMBER

ABSTRACT TITLE, PRESENTING AUTHOR/FIRST AUTHOR (if different from presenting)

- | | |
|----|--|
| 1 | Modelling FSHD by cell transplantation and genetic manipulation in a zebrafish model.
<i>Camilla Farnetani</i> |
| 2 | The role of estrogen receptor related beta (ESRR β) in FSHD-1 mechanism.
<i>Anna Pakula</i> |
| 3 | U7-snRNA-Mediated Exon Skipping of the Toxic DUX4 Gene as a Promising Therapeutic Approach for Facioscapulohumeral Muscular Dystrophy. (Corrected.)
<i>Afroz Rashnonejad</i> |
| 4 | The RNA binding protein FRG1 controls transcription landscape regulating muscle maturation and metabolism.
<i>Antonio Vallarola</i> |
| 5 | Circulating biomarkers for facioscapulohumeral muscular dystrophy.
<i>Yi-Wen Chen, Christopher R Heier</i> |
| 6 | Measurement properties of performance-based outcome measures of physical functioning in individuals with facioscapulohumeral dystrophy (FSHD) - A systematic review.
<i>Katy de Valle</i> |
| 7 | Process Abstract: Implementing Kinect sensor-based reachable workspace (RWS) measurement system in a multi-site, international FSHD clinical study.
<i>Jay Han</i> |
| 8 | Proposal of a neuropsychological protocol to study cognitive functions in FSHD.
<i>Elisa Lai</i> |
| 9 | Self-report questionnaire vs. clinical evaluation form in the French National Registry on facioscapulohumeral dystrophy: a statistical comparison.
<i>Benoit Sanson</i> |
| 10 | Patterns of muscle involvement, predictive characteristics, and meaningful change for functional motor tasks in FSHD.
<i>Jeffrey M Statland</i> |
| 11 | Biomarker identification by high-resolution proteomic approach in FSHD.
<i>Giorgio Tasca, Victor Corasolla Carregari</i> |
| 12 | Effect of Creatine Monohydrate on Functional Muscle Strength and Muscle Mass in Children with FSHD: a Multi-Centre, Randomised, Double-Blind Placebo-Controlled Crossover Trial
<i>Ian Woodcock</i> |
| 13 | Phenotype may predict the clinical severity of facioscapulohumeral muscular dystrophy.
<i>Wenhua Zhu, Yiqi Liu</i> |
| 14 | An in situ hybridization-based method for detecting DUX4 RNA expression in vitro.
<i>Amini Chermahini-Gholamhossein</i> |
| 15 | Characterization of human perivascular cells as a new cellular model of facioscapulohumeral muscular dystrophy.
<i>Giorgia di Blasio</i> |
| 16 | Investigation of the effect of estrogen on DUX4/ β -catenin/PAX3-7 protein levels in facioscapulohumeral muscular dystrophy (FSHD).
<i>Ceren Hangül</i> |
| 17 | Single-cell transcriptomics reveals DUX4 expression during early stages of myogenesis in FSHD1.
<i>Oliver King, Anna Pakula</i> |



POSTER SESSIONS

PALACE DU PHARO, GRAND LARGE (1st level)

Odd-numbered posters on July 19, 18:00-20:00

Even-numbered posters on July 20, 12:45-14:30

POSTER NUMBER

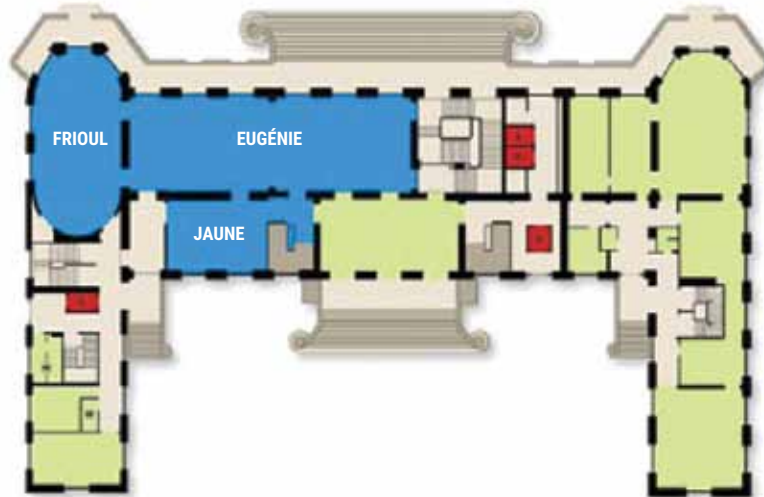
ABSTRACT TITLE, PRESENTING AUTHOR/FIRST AUTHOR (if different from presenting)

- 18** Exploring the relationship between DUX4 and hypoxia-inducible factor (HIF1 α).
Thuy-Hang Nguyen
- 19** DUX4 is a co-repressor of the progesterone and glucocorticoid nuclear hormone receptor.
Alberto Luis Rosa, Julieta Quintero
- 20** Deciphering the mechanism of herpesviral DUX4 induction.
Stephanie Walter
- 21** Genetic and epigenetic analysis of the FSHD-linked 4q35 region in rare female "Coats' disease" patients.
Robin Fitzsimons
- 22** SMCHD1 plays a pleiotropic role in euchromatin or heterochromatin regulation with consequences in rare diseases.
Camille Laberthonnière, Camille Dion
- 23** NO66 acts together with SMCHD1 as a co-repressor of DUX4 in facioscapulohumeral muscular dystrophy (FSHD).
Mara Tihaya
- 24** Do different phenotypes predict the clinical course of FSHD?
Giulia Ricci
- 25** An innate immunity model of FSHD muscle pathology.
Katelyn Daman
- 26** Elucidating the role of metabolic stress and mitochondrial dysfunction in FSHD.
Philipp Heher
- 27** Intramuscular pattern of fat infiltration measured by MRI to identify disease initiation in FSHD.
Linda Heskamp
- 28** The French National Registry of patients with facioscapulohumeral muscular dystrophy.
Céline Guien
- 29** An update on the UK FSHD Patient Registry in 2019 and future considerations.
Ben Cody Porter
- 30** Safety and tolerability of losmapimod, a selective p38/MAPK inhibitor, for treatment of FSHD at its root cause.
Diego Cadavid
- 31** Antisense therapy for facioscapulohumeral muscular dystrophy.
Yi-Wen Chen, Aiping Zhang
- 32** Phase 1 clinical trial of losmapimod in FSHD.
Michelle L Mellion
- 33** Losmapimod reduces DUX4 expression across FSHD patient-derived myotube cells.
Alejandro Rojas
- 34** MyoScreen™, a drug discovery platform for FSH muscular dystrophy.
Joanne Young
- 35** Single-nucleus RNA-seq identifies divergent populations of FSHD2 myotube nuclei.
Katherine Williams, Shan Jiang

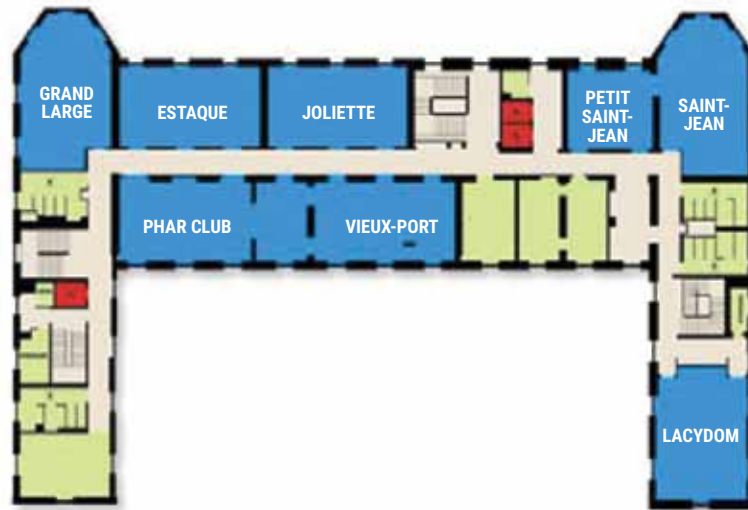


FLOOR MAPS

GROUND LEVEL



FIRST LEVEL



SECOND LEVEL



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