Facioscapulohumeral Dystrophy: From the clinics to the bench

Organizing body: FSH Society, June Kinoshita, Chief Strategic Programs Officer

Host organization: Marseille Medical Genetics Center; Aix Marseille Université, INSERM; GIPTIS: Genetic Institute for Patients, Therapies and Innovative Science; Department of Medical Genetics, La Timone Children’s Hospital, Marseille, FRANCE.

Co-chairs: Frédérique Magdinier, PhD, and George Padberg, MD.

Program Committee: Alexandra Belayew, PhD, Sabrina Sacconi, MD PhD, Stephen Tapscott, MD PhD, Rabi Tawil, MD, Rossella Tupler, MD PhD, Peter Zammit, PhD

Overview
The 2019 FSHD International Research Congress will bring together clinicians, scientists, patient representatives, and policy makers to discuss the latest developments in facioscapulohumeral muscular dystrophy (FSHD). Since 1996, this workshop has provided the FSHD community with a forum to present and discuss new developments, reinforce collaborative efforts, facilitate new initiatives, and coordinate research and clinical activities. With the recent advances in FSHD studies, the numerous developments, large sets of data, greater availability of biomaterials from multiple large research initiatives, a meeting for dissemination of new ideas, discussion of new strategies, collaboration and coordination is highly needed. The conference also presents opportunities for patient advocacy groups to participate and meet.

Our generous sponsors
Keynote Speaker Profiles

Michel Fardeau. Professor Fardeau is Emeritus Research Director at the 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 Français contre les Myopathies.

Bradley R. Cairns, PhD. 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 he 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 of 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.
Preliminary Program

Day 1

7h30-8h30  Registration. *Please take breakfast at your hotel before coming to the Pharo Palace.*

8h30-8h35  Welcome
Mark Stone, President & CEO
George Padberg & Frédérique Magdinier, Co-chairs

8h35-9h00  Plenary: FSHD, the patients’ perspective

9h00-9h30  Keynote: The FSHD phenotype and genotype
Prof. Michel Fardeau, Institute of Myologie
Understanding the complexity in the FSHD phenotype and variability in term of phenotype and genotype is a key point for all aspects related to the disease, from basic research, registries, or development of cures.

9h30-9h45  Report on the March 2019 Industry Collaborative Workshop on FSHD Therapeutics
Jamshid Arjomand, PhD, Chief Science Officer, FSH Society

9h45-10h10  Coffee break

10h10-11h10  Session 1. The FSHD clinical phenotype *(4 talks, 60 minutes)*
Chair/co-moderator George Padberg, MD

11h10-12h10  Session 2, Genetics and molecular findings for genotype-phenotype correlations and genetic diagnostics *(4 talks, 60 minutes)*
Chair/co-moderator Rossella Tupler, MD PhD

12h10-13h15  Lunch.

13h15-15h00  Session 3. Molecular mechanisms: DUX4, downstream targets, other players *(7 talks, 105 minutes)*
Chair/co-moderator Pete Zammit

15h00-15h30  Coffee break

15h30-17h00  Session 4. Molecular mechanisms and animal models *(6 talks, 90 minutes)*
Chair/co-moderator Alexandra Belayew, PhD

17h00-18h00  Break

18h00-20:00:  Cocktails + poster session

20h00-22h30  Conference Banquet
Pharo Palace
Day 2

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

8h25-8h30  Welcome
George Padberg & Frédérique Magdinier, Co-chairs

8h30-9h15  Keynote: The role of DUX4 in development
Brad Cairns, PhD, University of Utah

9h15-10h15  Session 5. DNA methylation and Epigenetics (4 talks, 60 minutes)
Chair/co-moderator Frédérique Magdinier, PhD

10h15-10h30  Coffee break

10h30-11h30  Session 6. Muscle pathology and imaging (4 talks, 60 minutes)
Chair/co-moderator Rabi Tawil, MD

11h30-12h15  Session 7. Natural history (3 talks, 45 minutes)
Chair/co-moderator Sabrina Sacconi, MD PhD

12h15-12h45  Session 8. Report on National Registries (1 talk, 30 minutes)
Karlien Mul, MD PhD

12h45-14h30  Lunch break + Poster session

14h30-16h00  Session 9. Therapeutic interventions (6 talks, 90 minutes)
Chair/co-moderator Stephen J. Tapscott, MD PhD

16h00-17h00  Session 10. Clinical evaluation, outcome measures, clinical trial readiness (4 talks, 60 minutes)
Chair/co-moderator Jeffrey Statland, MD

17h00-17h15  Coffee break

17h15-18h00  Session 11. Meeting conclusion, priorities discussion.
Moderated discussion to synthesize the meeting presentations, review priorities for the FSHD field in basic and translational research and clinical trial readiness.

18h00  Adjourn