



fsrmm

fondation suisse de recherche sur les maladies musculaires
fondazione svizzera per la ricerca sulle malattie muscolari
schweiz. stiftung für die erforschung der muskelkrankheiten

Program

14th Swiss Meeting on Muscle Research

Macolin / Magglingen

3rd – 5th December 2023



Vesalius (1514-64)

Sunday, December 3rd

16:00-17:30 Arrival, Check-in

17:30-18:30 Welcome Aperó

18:30-19:30 Dinner

19:30-19:35 Meeting opening (Markus A. Rüegg)

19:35-20:20 Keynote lecture: Challenges in ALS: from diagnosis to drug development

Special guest: Markus Weber, Neuromuscular centre/ALS clinic, Kantonsspital St. Gallen

Session 1: New developments for ALS

Chair: Markus Rüegg

20:20-20:50 Sphingolipids as emerging players in amyotrophic lateral sclerosis

Museer Lone, University Hospital Zürich

20:50-21:20 Precision amidst dynamic chaos: Unveiling moving targets of ALS

Leonidas Emmanouilidis, ETH Zürich

Monday, December 4th

Session 1: New developments for NMDs

Chair: Susan Treves

8:30-9:00 Pathomechanisms involved in the selective affection of skeletal muscle in X-linked

Myopathy with Excessive Autophagy

Ilaria Cocchiararo, University of Geneva

9:00-9:30 RyR1 content decrease-induced ER stress is a hallmark of myopathies

Nadège Zanou, University of Lausanne

9:30-10:00 AAV-mediated therapeutic effect of linker protein-mediated gene therapy on muscle and nerve pathology in mouse models for LAMA2 MD

Judith Reinhard, University of Basel

10:00-10:30 Uncovering Wif1 contribution to muscular dystrophy phenotypes

Eleonora Maino, University of Basel

10:30-11:00 Coffee break

11:00-11:30 Interaction between the Ca²⁺ channel Orai3 and the scaffold protein AHNAK2:
implication during muscle stem cell activation
Stephane Koenig, University of Geneva

11:30-12:00 Store Operated Calcium Entry regulates synaptic gene expression and muscle response
to denervation
Alexandre Prola, University of Geneva

12:00-12:30 Exploiting S-acylation of STIM proteins in muscle
Amado Carreras Sureda, University of Geneva

12:30-13:30 Lunch

Poster session

13:30-15:30 ALL POSTERS, *coffee and tea*

Session 3: Muscle function**Chair: Thomas Laumonier**

15:30-16:00 Biofabrication of a 3D microphysiological model of human skeletal muscle for the study
of fibrosis in aging and disease
Simone Bersini, University of Lugano

16:00-16:30 The neuromuscular junction: the chink in skeletal muscle's armor in neuromuscular
pathologies and other diseases
Christoph Handschin, University of Basel

16:30-17:00 New aspects of TGFβ signaling in muscle regeneration
Jérémy Kessler, University of Geneva

17:00-17:30 Exploring the multifaceted signaling in skeletal muscle of geriatric mice
Daniel Ham, University of Basel

17:30-17:50 Break

Session 4: Stem cells**Chair: Frédéric Allain**

17:50-18:20 Interspecies generation of iPSC-derived functional muscle stem cells
Ori Bar-Nur, ETH Zürich

18:20-18:50 Uncovering a novel myogenic regulator that facilitates reprogramming of fibroblasts into
functional myogenic progenitor cells
Giada Bacchin, ETH Zürich

18:50-19:20 Intracellular autofluorescence allows the isolation of functional human MuRC subpopulations with distinct cell states
Axelle Bouche, University of Geneva

19:30-20:30 Dinner
Poster prize committee meets over dinner

Evening program

20:30-23:00 free beer at poster site

Tuesday, December 5th

Session 1: Mitochondrial involvement in disease mechanisms

Chair: Maud Frieden

9:00-9:30 Integrated nutrient-sensitized screens predict mitochondrial disease genes
Alexis Jourdain, University of Lausanne

9:30-10:00 LACTB, a new player in lipid modulation
Sylvaine Lagarrigue, University of Lausanne

10:00-10:30 MitoBooster: a novel strategy for Inducing mitophagy and enhancing cellular homeostasis
Ammar Ebrahimi, University of Lausanne

10:30-10:45 Concluding remarks and poster prizes (Markus Rüegg)

10:45-11:15 Coffee Break and Poster removal

DEPARTURE

Poster presentations

Author	Title	Nr
New findings for neuromuscular diseases		
Imam Rostami	Structural analysis of synapse impairment in cells derived from ALS patients with C9ORF72 mutation using cryo-electron tomography	1
Daria Barbash	Investigation of SRSF1 interactions with GC-rich RNA repeats and consequences on phase separation in the context of ALS/FTD	2
Rim Diab	Selective impairment of autophagy in C9ORF72-linked ALS	3
Nicole Ziak	Understanding the selective neurotoxicity of serine-palmitoyltransferase (SPT) mutations towards motor and sensory neurons	4
Sofia Benucci	Characterization of mouse model knocked in for the RyR1 p.F4976L mutation identified in a severely affected child: physiological, biochemical and structural features	5
Huiying Li	Functional consequences of MH-causative RYR1 mutations on the immune system	6
Hervé Meier	Genotypic and transcriptomic characterization of patients with King-Denborough Syndrome	7
Alexis Ruiz	Effect of RYR1 mutations on muscle spindle function and their impact on the musculoskeletal system	8
Samuele Metti	ANTXR2 regulates Collagen VI homeostasis in skeletal muscle	9
Regula Furrer	Metabolic dysregulation contributes to the development of dysferlinopathy	10
Sebastian Holdermann	Molecular mechanisms of complement activation and neuromuscular disruption by combinations of autoantibodies from patients with Myasthenia Gravis	11
Daehong Kim	Integrative analysis of the single nucleotide polymorphism and inflammatory cytokine responses in patients with Myasthenia Gravis	12
Magdalena Mroczek	Three heterozygous GAA cases mimicking late-onset Pompe disease	13
Loris Levet	Acute modulation of VMA21 leads to skeletal muscle degeneration	14
Sana Kouba	Studying the role of S-Acylation of ORAI1 channel and STIM1 protein in tubular aggregate myopathies	15
Dirk Fischer	Fumaric acid in adults with Becker muscular dystrophy: A single-centre, open-label, safety, and efficacy proof of concept 28-days trial	16
Aouatef Djeraba Ait-Lounis	The masseter muscle, an often-neglected muscle in Duchenne muscular dystrophy: a pilot study using mouse models	17
Julia Schedel	Investigating the Myotendinous Junction in the context of LAMA2-related Muscular Dystrophy	18
Alok Behera	LIN28 inhibition reverses miRNA expression and disease hallmarks in myotonic dystrophy	19
New findings in muscle function and plasticity		

Christine Trautmann	Cultivated Meat	20
Cassandra Tabasso	"Oh my DAG", unveiling the specific link between human insulin resistance and lipotoxicity	21
Axel Aguetaz	A novel PLIN3 splicing variant reveals a conserved mitochondrial targeting of perilipin protein family members	22
Shivani Mansingh	More than the clock: distinct regulation of muscle function and metabolism by PER2 and ROR α	23
Loann Laubry	STIM1 and STIM1L in skeletal muscle: central regulators of calcium circuitry	24
Mélanie Fourgeaud	Orai3 and its partner AHNAK2 regulate the activation of human skeletal muscle stem cells in vitro	25
Olivier Dupont	Roles of the two STIM2 isoforms in human myotube formation and function	26
Marina Baraldo	From nerve to muscle and vice versa: exploring how exercise shapes the motor unit and muscle-nerve communication	27
Lionel Tintignac	Molecular mechanisms modulating muscle protein synthesis	28