

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

# **Program**

11<sup>th</sup> Swiss Meeting on Muscle Research

Macolin / Magglingen

20<sup>th</sup> – 22<sup>th</sup> November 2016



Vesalius (1514-64)

# Sunday, November 20th

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

17:30-18:30 Welcome Apero

18:30-19:30 Dinner

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

19:35-20:20 Making sense of heterogeneities in adult skeletal muscle stem cells

<u>Special guest</u>: Shahragim Tajbakhsh, Institut Pasteur, Paris

#### **Session 1: Muscle Development**

20:20-20:50 Human myogenic reserve cells contribute to muscle regeneration and satellite cell formation after intramuscular transplantation in immunodeficient mice

Thomas Laumonier, University of Geneva

Chair: Markus A. Rüegg

- 20:50-21:20 Receptor FgfrL1 and slow muscle fibers Beat Trueb, University of Bern
- 21:20-21:50 Ablation of raptor impairs but does not abolish myogenesis during mouse development Nathalie Rion, Biozentrum, Basel

## **Sponsors 2016**





# Monday, November 21th

**Chair: Nicolas Mermod** 

**Chair: Laurent Bernheim** 

### **Session 1: Cell therapy approaches**

8:00-8:30	tba Jerome Feige, EPF Lausanne
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8:30-9:00	Role of NFAT proteins in human primary myoblast differentiation
	Julie Perroud, University of Geneva
9:00-9:30	NAD+ repletion improves mitochondrial and stem cell function and enhances life span in
	mice
	Hongbo Zhang, EPF Lausanne
9:30-10:00	Mesoangioblasts from different tissues: potential for muscle regeneration and
	translational perspective
	Marisa Jaconi, University of Geneva

### 10:00-10:45 Coffee break

### **Session 2: Disease-specific therapy approaches**

- 10:45-11:15 Deciphering the pathogenic mechanisms of C9ORF72 ALS and FTD Magdalini Polymenidou, University of Zürich
- 11:15-11:45 RNA binding proteins involved in neurodegeneration and neuro-muscular diseases Frederic Allain, ETH Zürich
- 11:45-12:15 The use of a mutated version of the splicing regulator SRSF1 (ASF/SF2) as a new strategy to cure Spinal Muscular Atrophy (SMA)

  Antoine Clery, ETH Zürich

#### 12:15-13:45 Lunch

- 13:45-14:15 Restoration of basement membrane assembly by small linker proteins prevents laminin- $\alpha 2$ -deficient muscular dystrophy Judith Reinhard, Biozentrum, Basel
- 14:15-14:45 Targeting deregulated AMPK and mTORC1 pathways in DM1 improves muscle function via splicing-dependent and -independent mechanisms

  Marielle Brockhoff, University of Basel

#### Poster session 1

14:45-16:30 POSTERS Nr. 1-13 and coffee break

- 16:30-17:00 U7 snRNA-based splicing modulation as a potential therapy for inherited diseases retrospective and outlook

  Daniel Schümperli, University of Bern
- 17:00-17:30 The Omigapil CALLISTO Studie in CMD patients Rudolf Hausmann, Santhera Pharmaceuticals
- 17:30-18:00 Enhancing estrogenic signalling to fight muscular dystrophies: Mechanisms of action and repurposing clinically approved drugs

  Elinam Gayi, University of Geneva
- 18:00-18:30 Rimeporide restores resting pH and decreases store operated calcium entry in dystrophic myotubes by inhibiting sodium hydrogen exchanger type 1 (NHE 1). Hesham Ismail, University of Geneva
- 18:30-19:00 Treatment with L-citrulline and metformin in Duchenne muscular dystrophy Dirk Fischer, University Hospital Basel

19:00-20.30 Dinner

#### Poster session 2

20:30-22:30 Posters and beer

## Tuesday, November 22th

**Chair: Michael Sinnreich** 

**Chair: Denis Monard** 

**Chair: Susan Treves** 

### **Session 1: Diagnostic tools**

8:30-9:00 Synchronous MRI of muscle motion induced by electrical stimulation Francesco Santini, University Hospital Basel

9:00-9:30 Neurophysiological assessment of muscle membrane properties Werner Z'Graggen, Inselspital Bern

9:30-10:15 Coffee Break

#### Session 2: Muscle plasticity and physiology (1)

10:15-10:45 Molecular mechanisms of HIIT-induced RyR1 modifications and skeletal muscle adaptations to exercise

Nadege Zanou, University of Lausanne

10:45-11:15 Extraocular muscles - RYR3: Is there something behind the eyes?

Jan Eckhardt, University Hospital Basel

11:15-11:45 Characterization of a transgenic mouse over-expressing SRP35 in its skeletal muscle: metabolic effect

Alexis Ruiz, University Hospital Basel

11:45-12:15 mTORC1 deregulation impairs muscle homeostasis upon denervation Perrine Castets, Biozentrum Basel

12:15-13:45 Lunch

#### Poster session 3

13:45-15:30 POSTERS Nr. 14-23

#### Session 3: Muscle plasticity and physiology (2)

15:30-16:00 Differential cytoplasmic adapter recruitment controls dynamic and function of beta-1 integrin splice variants

Bernhard Wehrle-Haller, University of Geneva

16:00-16:30 Urolithin A induces mitophagy and prolongs lifespan in C. elegans and increases muscle function in rodents

Laurent Mouchiroud, EPF Lausanne

16:30-17:00 The coactivator PGC-1alpha regulates skeletal muscle cell plasticity in health and disease Christoph Handschin, Biozentrum Basel

Concluding remarks (Markus Rüegg)

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Departure

# **Poster presentations**

Author	Title	Topic	Nr
Lei Zhuang	Role of the receptor FGFRL1 in muscle development	Muscle development	1
Pavithra Iyer	Assessment of a cell therapy approach for Duchenne muscular dystrophy using mesoangioblasts and non-viral vectors	Cell therapy	2
Omid Mashinchian	Derivation of uncommitted human muscle stem cells from iPSCs	Cell therapy	3
Sonia Karaz	Cross-talk between regeneration and ectopic adipogenesis in muscle aging	Cell therapy	4
Christoph Bachmann	Characterization of Excitation-Contraction Coupling in Muscles from Patients with X-Linked Myotubular Myopathy	Disease-specific approaches	5
Regula Furrer	Effect of exercise and exercise factors on cancer cachexia	Disease-specific approaches	6
Sophie Sauc	Differential roles of STIM1 and STIM1L in skeletal muscle	Disease-specific approaches	7
Olivier Dorchies	The mdx5Cv dystrophic mouse: first in depth longitudinal phenotyping	Disease-specific approaches	8
Hesham Hamed	Lack of TRPC1 cation channels enhances muscle fatigue in mdx5Cv dystrophic mice	Disease-specific approaches	9
Jochen Kinter	Identification of Plant-derived Alkaloids with Therapeutic Potential for Myotonic Dystrophy Type I	Disease-specific approaches	10
Nicolas Place	Neuromuscular adjustments in plantar flexors in young adults with cerebral palsy	Disease-specific approaches	11
Ryu Dongryeol	NAD+ repletion improves muscle function in muscular dystrophy and counters global PARylation.	Disease-specific approaches	12
Maurizio Sury	Attempt at further ameliorating the muscular dystrophy in mouse models for MDC1A using a construct combining "mini-agrin" and "aLNNdÂ	Disease-specific approaches	13
Olivier Dorchies	Estrogen deprivation caused by the lack of aromatase aggravates the pathology in dystrophic mice	Translational medicine	14
Beate Rücker	Cardiac involvement in patients with Duchenne / Becker muscular dystrophy (Duchenne heart study) – a longitudinal observational study	Diagnostic tools	15
Xeni Deligianni	Reproducibility of EMS-induced synchronous MRI of muscle motion	Diagnostic tools	16
Kathrin Chojnowska	Regulation of the mitochondrial dynamics via mTORC1 in skeletal muscle	Muscle plasticity and physiology	17

Maud Frieden	Ion channel(s) gated by STIM1L, an isoform of STIM1 highly expressed in skeletal muscle	Muscle plasticity and physiology	18
Laurence Neff	PCR-restriction-based strategies allow genotyping without sequencing of several allelic variants of the mdx mouse that carry point mutations	Muscle plasticity and physiology	19
Laurence Neff	Towards generating new dystrophic mouse mutants by homologous recombination using dimeric RNA-guided Fokl-dCas9 nucleases	Muscle plasticity and physiology	20
Jonathan Gill	PGC-1a amelioration of mitochondria function and calcium metabolism prevents age-related formation of tubular aggregates in muscle	Muscle plasticity and physiology	21
Shuo Lin	Sustained activation of mTORC1 affects the integrity and function of the neuromuscular junction reminiscent of age-related changes	Muscle plasticity and physiology	22
Lionel Tintignac	Deregulation of the proteostasis network in diseased muscle	Muscle plasticity and physiology	23