

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>nd</sup> November 2016



Vesalius (1514-64)

Organizer: Prof. Markus A. Rüegg, Biozentrum, University of Basel

### 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

Special guest: 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**

KontaktGruppe für Forschungsfragen
Contact Group for Research Matters



### Monday, November 21st

Chair: Denis Monard

**Chair: Laurent Bernheim** 

Session 1. Cen therapy approaches		Chair. Dellis Worlard
8:00-8:30	Targeting the stem cell niche to restore muscle regene Jérôme Feige, EPF Lausanne	eration in aging
8:30-9:00	Role of NFAT proteins in human primary myoblast diff Julie Perroud, University of Geneva	erentiation
9:00-9:30	NAD+ repletion improves mitochondrial and stem cell mice Hongbo Zhang, EPF Lausanne	function and enhances life span in
9:30-10:00	Mesoangioblasts from different tissues: potential for r translational perspective	nuscle regeneration and

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

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

Marisa Jaconi, University of Geneva

Session 1: Cell 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 Frédéric 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. 1-16 and coffee break

- 16:30-17:00 Cardiac involvement in patients with Duchenne / Becker muscular dystrophy (Duchenne heart study) a longitudinal observational study

  Beate Rücker, Kinderspital Zürich
- 17:00-17:30 Synchronous MRI of muscle motion induced by electrical stimulation Francesco Santini, University Hospital Basel
- 17:30-18:00 Neurophysiological assessment of muscle membrane properties Werner Z'Graggen, Inselspital Bern
- 18:00-18:30 Urolithin A induces mitophagy and prolongs lifespan in C. elegans and increases muscle function in rodents

  Laurent Mouchiroud, EPF Lausanne

19:00-20.30 Dinner

#### **Evening program**

20:30-22:30 free beer at poster site

## Tuesday, November 22nd

Session 1: Translational medicine		Chair: Urs Ruegg
8:00-8:30	U7 snRNA-based splicing modulation as a potential therapy for in retrospective and outlook Daniel Schümperli, University of Bern	nherited diseases -
8:30-9:00	The Omigapil CALLISTO Studie in CMD patients Rudolf Hausmann, Santhera Pharmaceuticals	
9:00-9:30	Enhancing estrogenic signalling to fight muscular dystrophies: M repurposing clinically approved drugs Elinam Gayi, University of Geneva	echanisms of action and
9:30-10:00	Therapeutic effectiveness of Rimeporide, an NHE-1 inhibitor, fro to models of Duchenne muscular dystrophy Hesham I.Hamed, University of Geneva	m muscle cells in culture
10:00-10:30	Treatment with L-citrulline and metformin in Duchenne muscula Dirk Fischer, University Hospital Basel	r dystrophy

#### 10:30-11:15 Coffee Break

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

- 11:15-11:45 mTORC1 deregulation impairs muscle homeostasis upon denervation Perrine Castets, Biozentrum Basel
- 11:45-12:15 Molecular mechanisms of HIIT-induced RyR1 modifications and skeletal muscle adaptations to exercise

  Nadège Zanou, University of Lausanne
- 12:15-12:45 Extraocular muscles RYR3: Is there something behind the eyes?

  Jan Eckhardt, University Hospital Basel

12:45-14:00 Lunch

#### Poster session 2

14:00-15:30 POSTERS Nr. 17-26

**Chair: Nicolas Mermod** 

- 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 The coactivator PGC-1alpha regulates skeletal muscle cell plasticity in health and disease Christoph Handschin, Biozentrum Basel
- 16:30-17:00 Characterization of a transgenic mouse over-expressing SRP35 in its skeletal muscle: metabolic effect

  Alexis Ruiz, University Hospital Basel

Concluding remarks (Markus Rüegg)

-please remove posters now-

Departure

# **Poster presentations**

Author	Title	Topic	Nr
Lei Zhuang	Role of the receptor FGFRL1 in muscle development	Muscle development	1
Stéphane König	Role of secreted pla2g5 during differentiation of DMD myoblast	Muscle development	2
Shilpy Joshi	Differential contributions of TEAD transcription factors to myogenic differentiation of C2C12 cells and primary myoblasts	Muscle development	3
Peter Meister	Antagonistic roles of Polycomb repression and Notch signaling impair muscle transdifferentiation in C. elegans	Muscle development	4
Pavithra lyer	Assessment of a cell therapy approach for Duchenne muscular dystrophy using mesoangioblasts and non-viral vectors	Cell therapy	5
Omid Mashinchian	Derivation of uncommitted human muscle stem cells from iPSCs	Cell therapy	6
Sonia Karaz	Cross-talk between regeneration and ectopic adipogenesis in muscle aging	Cell therapy	7
Christoph Bachmann	Characterization of Excitation-Contraction Coupling in Muscles from Patients with X-Linked Myotubular Myopathy	Disease-specific approaches	8
Sophie Saüc	Differential roles of STIM1 and STIM1L in skeletal muscle	Disease-specific approaches	9
Olivier Dorchies	The mdx5Cv dystrophic mouse: first in depth longitudinal phenotyping	Disease-specific approaches	10
Hesham Hamed	Lack of TRPC1 cation channels enhances muscle fatigue in mdx5Cv dystrophic mice	Disease-specific approaches	11
Jochen Kinter	Identification of plant-derived alkaloids with therapeutic potential for Myotonic Dystrophy Type I	Disease-specific approaches	12
Nicolas Place	Neuromuscular adjustments in plantar flexors in young adults with cerebral palsy	Disease-specific approaches	13
Dongryeol Ryu	NAD+ repletion improves muscle function in muscular dystrophy and counters global PARylation.	Disease-specific approaches	14
Maurizio Sury	Attempt at further ameliorating the muscular dystrophy in mouse models for MDC1A using a construct combining "mini-agrin" and "alnnd"	Disease-specific approaches	15
Ruben Herrendorff	A novel antigen-specific treatment for multifocal motor neuropathy	Disease-specific approaches	16
Xeni Deligianni	Reproducibility of EMS-induced synchronous MRI of muscle motion	Diagnostic tools	17

Olivier Dorchies	Estrogen deprivation caused by the lack of	Translational	18
Olivier Dorchies	aromatase aggravates the pathology in dystrophic mice	medicine	
Kathrin Chojnowska	Regulation of the mitochondrial dynamics via mTORC1 in skeletal muscle	Muscle plasticity and	19
Rathini Chojnowska		physiology	
Maud Frieden	Ion channel(s) gated by STIM1L, an isoform of STIM1	Muscle plasticity and	20
	highly expressed in skeletal muscle	physiology	
Laurence Neff	PCR-restriction-based strategies allow genotyping	Muscle plasticity and	21
Laurence Nen	without sequencing of several allelic variants of the mdx mouse that carry point mutations	physiology	
Laurence Neff	Towards generating new dystrophic mouse mutants	Muscle plasticity and	22
Laurence Neir	by homologous recombination using dimeric RNA-	physiology	
	guided Fokl-dCas9 nucleases		
Jonathan Gill	PGC-1a amelioration of mitochondrial function and	Muscle plasticity and	23
	calcium metabolism prevents age-related formation	physiology	
	of tubular aggregates in muscle	NA la . la . l'all l	2.4
Shuo Lin	Sustained activation of mTORC1 affects the integrity	Muscle plasticity and	24
	and function of the neuromuscular junction reminiscent of age-related changes	physiology	
Dogula Furrar	PGC-1α-induced cross-talk between muscles and	Muscle plasticity and	25
Regula Furrer	macrophages may prime muscles for faster regeneration	physiology	
11 1 <del>- 1</del> 1		Muscle plasticity and	26
Lionel Tintignac	Deregulation of the proteostasis network in diseased muscle	physiology	
Francesco Zorzato	An RYR1 mutation associated with malignant	Muscle plasticity and	27
Francesco Zorzato	hyperthermia is also associated with bleeding abnormalities	physiology	