

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

10th Swiss Meeting on Muscle Research Macolin / Magglingen 9th – 11th November 2014



Vesalius (1514-64)

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

Sunday, November 9th

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

17:30-18:30 Welcome Apero

18:30-20:00 Dinner

20:00-20:10 Meeting opening (Markus A. Rüegg)

20:10-20:40 Rare conditions and global thinking - the impact of international networking Special guest: Marita Pohlschmidt, Muscular Dystrophy Campaign, UK

Session 1: New targets for DMD and SMA

20:40-21:10 Structural and functional investigations reveal promising targets to cure Spinal Muscular Atrophy

Antoine Clery, ETH Zürich

21:10-21:40 Enhancing estrogenic signalling to fight muscular dystrophies: Mechanisms of action and repurposing clinically approved drugs

Olivier Dorchies, University of Geneva

Sponsors 2014





Chair: Markus A. Rüegg





Monday, November 10th

| 8:20-8:50 | Key role of Epidermal Growth Factor Receptor during human primary myoblast differentiation Julie Perroud, University of Geneva | | | |
|---|--|----------------------|--|--|
| 8:50-9:20 | Characterization of the trafficking and functional properties of the STIM1 isoform Sophie Saüc, University of Geneva | muscle specific long | | |
| 9:20-9:50 | The regulation of skeletal muscle cell plasticity by PGC-1alpha in health and disease Christoph Handschin, Biozentrum, Basel | | | |
| 9:50-10:20 | Identification of new regulators of muscle size and ageing Lionel Tintignac, University of Basel | | | |
| | 10:20-10:40 Coffee break | | | |
| Session 2: Cell therapy | | Chair: Urs Rüegg | | |
| 10:40-11:00 Improving cellular therapies of muscle dystrophies by uncovering epigenetic and signaling pathways of muscle formation Peter Meister, University of Bern | | | | |
| 11:00-11:30 Muscle gene transfer mediated by mesoangioblasts and non-viral vector systems | | | | |

11:30-12:00 VEGF gene transfer enhances human muscle derived stem cells survival after transplantation in injured skeletal muscles

Thomas Laumonier, University Hospital, Geneva

Pavitra Iyer, University of Lausanne

12:00-13:10 Lunch

Poster session 1

13:15-15:30 POSTERS Nr. 1-21

Session 1: Muscle regeneration and plasticity

Session 3: Work with patients

Chair: Laurent Bernheim

Chair: Denis Monard

15:30-16:00 Efficacy of idebenone on respiratory outcome in DMD patients not using glucocorticoid steroids: Results of a phase III double-blind, randomised, placebo-controlled multicenter trial

Thomas Meier, Santhera Pharmaceuticals, Liestal

- 16:00-16:30 L-citrulline and metformin trials in Duchenne and Becker muscular dystrophy Dirk Fischer, University Hospital Basel
- 16:30-16:50 Participation of Swiss patients in the large multinational trials Andrea Klein, Kinderspital Zürich

16:50-17:20 Muscle velocity recovery cycles: as indicators of resting membrane potential Werner Z'Graggen, University Hospital Bern

17:20-17:40 Coffee Break

Chair: Nicolas Mermod

Session 4: New targets for ALS and LGMD2B

17:40-18:10 Marinesco-Sjögren syndrome protein SIL1 modulates motoneuron vulnerability to ER stress and degeneration in ALS Smita Saxena, University of Bern

18:10-18:40 Translational treatment approaches for muscular dystrophies Michael Sinnreich, Pharmazentrum, Basel

18.40-20.00 Dinner

Poster session 2

20:00-22:00 POSTERS Nr. 22-34

Tuesday, November 11th

Session 1: New pathways in muscle pathology (1) Chair: Matthias Chiquet

| 8:30-9:00 | Skeletal muscle mTORC1 activation alters global metabolism Maitea Guridi, Biozentrum, Basel |
|------------|---|
| 9:00-9:30 | Acetylation controls beta1A integrin localization and function Birgit Kastberger, University of Geneva |
| 9:30-10:00 | Cardiac sodium channel NaV1.5 distribution in myocytes via interacting proteins: the multiple pool model Ludovic Gillet, University of Bern |

10:00-10:30 Coffee Break

Session 2: New pathways in muscle pathology (2)

10:30-11:00 Slow muscle fibers gradually die by apoptosis in FgfrL1-deficient mice Beat Trueb, University of Bern

11:00-11:30 Enhanced glucose uptake in skeletal muscle of transgenic mice overexpressing SRP35 Susan Treves, University Hospital, Basel

11:30-12:00 Concluding remarks, Poster prize (Markus Rüegg)

-please remove posters now-

12:00-13:00 Lunch

Departure

Chair: Francesco Zorzato

Poster presentations

| Author | Title | Topic | Nr |
|------------------------|---|------------------------------------|----|
| Carlo Rossi | Cardiac regeneration potential of fetal mesoangioblasts from aorta, cardiac and skeletal muscle using engineered hyaluronic acid- or PEGbased hydrogels | Cell therapy | 1 |
| Francesca Coraggio | Improving cellular therapies of muscle dystrophies by uncovering epigenetic and signaling pathways of muscle formation | Cell therapy | 2 |
| Flavien Bermont | VEGF gene transfer enhances human muscle derived stem cells survival after transplantation in injured skeletal muscles | Cell therapy | 3 |
| Flavio_Ronzoni | Transcriptional signature of human mesoangioblasts from different fetal tissues | Cell therapy | 4 |
| Florian Bentzinger | Wnt7a stimulates myogenic stem cell motility and engraftment resulting in improved muscle strength | Cell therapy | 5 |
| Daniel Schümperli | Splicing correction for Erythropoietic Protoporphyria | Gene therapy | 6 |
| Philipp Odermatt | Uncovering the role of microRNAs in SMA | Gene therapy | 7 |
| Luca Borradori | Characterization of the function of the spectraplakin BPAG1 in myoblasts | Muscle regeneration and plasticity | 8 |
| Maud Frieden | Characterization of the trafficking and functional properties of the muscle specific long STIM1 isoform | Muscle regeneration and plasticity | 9 |
| Karima Habbout | Modulations of transcription factors during human primary myoblast differentiation. | Muscle regeneration and plasticity | 10 |
| Stephane Konig | Modulations of transcription factors during human primary myoblast differentiation. | Muscle regeneration and plasticity | 11 |
| Jonathan Gill | PGC-1α and Exercise: how to slow aging and sarcopenia | Muscle regeneration and plasticity | 12 |
| Lei Zhuang | The receptor FgfrL1 is specifically required for development and survival of slow muscle fibers | Muscle regeneration and plasticity | 13 |
| Laura Lukjanenko | Mechanisms of regeneration and ectopic adipogenesis during aging of skeletal muscle | Muscle regeneration and plasticity | 14 |
| Hesham Ismail Hamed | Diapocynin, a putative NADPH oxidase inhibitor, ameliorates the phenotype of a mouse model of Duchenne muscular dystrophy | New targets in NMD | 15 |
| Amparo Garcia-Lopez | Identifying small molecules targeting an RNA stem- loop involved in the alternative splicing of the SMN2 gene: a therapeutic target in SMA | New targets in NMD | 16 |

| Ruben Herrendorff | Identification of Small Molecules with Therapeutic Potential for Myotonic Dystrophy Type I | New targets in NMD | 17 |
|----------------------------|--|----------------------------------|----|
| Jochen Kinter | Therapeutic Strategy for Facio-Scapulo-Humeral Muscular Dystrophy using Aptamers | New targets in NMD | 18 |
| Anuja Neve | Functional consequences of spinal muscular atrophy at the neuromuscular junction | New targets for ALS | 19 |
| Céline Ruegsegger | Identification of the aberrant interactome of mutant SOD1 in familial amyotrophic lateral sclerosis model of disease | New targets for ALS | 20 |
| Niran Maharjan | Unravelling the role of a novel protein C9ORF72 in ALS and FTD | New targets for ALS | 21 |
| Nathalie Rion | Ablation of raptor, but not rictor, impairs myogenesis during mouse development | New pathways in muscle pathology | 22 |
| Marijana Sekulic | Characterization of excitation contraction coupling components in human extraocular muscles | New pathways in muscle pathology | 23 |
| Perrine Castets | Denervation worsens the myopathy related to active mTORC1 by affecting autophagy | New pathways in muscle pathology | 24 |
| Marielle Brockhoff | Deregulation of the mTORC1 signalling and impairment of the autophagy process as pathomechanisms for Myotonic Dystrophy type I | New pathways in muscle pathology | 25 |
| Ruben Lopez Dicuru | Excitation-contraction coupling is affected by Raptor ablation (mTORC1) in skeletal muscle | New pathways in muscle pathology | 26 |
| Isabelle Vögeli | Function, Pharmacology and Pathophysiology of Transient Receptor Potential Canonical Channel 1 (TRPC1) | New pathways in muscle pathology | 27 |
| Ori Rokach | Mechanism of action of recessive RYR1 mutations | New pathways in muscle pathology | 28 |
| Marco Kaiser | Meta-analysis of mTORC1-regulated transcriptional networks in skeletal muscle | New pathways in muscle pathology | 29 |
| Bernhard Wehrle- Haller | Modulation of integrin adhesion-signalling during muscle cell differentiation | New pathways in muscle pathology | 30 |
| Alexis Ruiz | SRP35 and glucose uptake, metabolic and Physiologic effect. | New pathways in muscle pathology | 31 |
| Shuo Lin | The role of Akt activation for the myopathy caused by sustained activation of mTORC1 | New pathways in muscle pathology | 32 |
| Tatiana Wiktorowicz | Novel mouse models for therapeutic studies of dysferlinopathies | New targets in NMD | 33 |
| Charlotte Lorin | Dystrophic cardiomyopathy - role of TRPV2 channels in stretch-induced cell damage | New pathways in muscle pathology | 34 |