



Annual Meeting of SGIEM

Thursday 22th April 2021



Source: MySwitzerland.com

*National Network and Scientific Symposium
of the SGIEM*

Final Program

General Information

Meeting Venue (Morning and Afternoon)

Inselspital Bern
Auditorium E. Rossi
Kinderklinik
Entrance 31b
3010 Bern

Tel: 031/632 45 52

Link for Online participation will be sent following individual registration

Organization

Swiss Group for Inborn Errors of Metabolism (SGIEM)
Universitätsklinik für Kinderheilkunde, INSELSPITAL
Freiburgstrasse 15
CH-3010 Bern

Steering Committee

Dr. med. Matthias Gautschi, Bern	Chairman
PD Dr. med. Diana Ballhausen, Lausanne	Secretary
PD Dr. med. Jean-Marc Nuoffer, Bern	Treasurer
Prof. Dr. med. Matthias Baumgartner, Zürich	
Dr. med. Ilse Kern, Genève	
PD Dr. med. Marianne Rohrbach	
Dr. med. Christel Tran, Lausanne	

Meeting Schedule

Morning (for details, see separate Agenda)

General meeting of the SGIEM, the National Metabolic Network

- 09:15 Opening General Meeting, Matthias Gautschi
- 09:30 General Meeting part 1
- 10:15 Coffee break
- 10:45 General Meeting part 2
- 11:45 Lunch break

Subgroup Meetings

12.15-13.10 Uhr

- Room "Kursraum 2" Dietician Group
- Clubraum Adult Group
- Auditorium E. Rossi SGBOND

Afternoon (open to all interested; on registration only)

Scientific Symposium

Venue: Auditorium Ettore Rossi, Kinderklinik, Entrance 31b AND/OR Online

13:15 KEYNOTE LECTURE: The Dos and Don'ts of Reference Centre Setup

The French experience by Sophie BERNICHTEIN, PhD, Paris

14:00-15:30 SESSION 1: Emerging Therapies – Chair: Diana Ballhausen

14:00 **Christel Tran:** ERT for late-onset M. Pompe disease

14:20 **New Therapies for Niemann-Pick Type C?**

Marianne Rohrbach: Cyclodextrin and Arimoclomol...

Tatiana Bremova-Ertl: N-Acetyl-DL-Leucine for NPC - Comparison of compounds, preclinical data & the IB1001 phase II clinical trial

15:00 **Experience of a patient family**

Anaïs Di Nardo: *Thérapie génique de la MPS IIIA - témoignage d'une maman (in French with English translation)*

15:20-15:45 Break

15:45-16:30 **SESSION 2: SGBOND News & Discussions** – Chair: Albrecht Popp

15:45 **Elena Gonzalez Rodriguez:** Burosumab treatment for XLH

16:00 **Alexandra Lenoir:** OI and respiratory problems – results from a study

16:15 **Cornelia Jungo:** Self-management in OI

16:30-17:15 SESSION 3: Updates & Research – Chair: Johannes Häberle

16:30 **Patrick Forny:** 1st Revision of Guidelines for MMA & PA

16:45 **Alina Castillo:** Mitochondrial Bioenergetics in human skin fibroblasts

17:00 **Mary Gonzalez:** The first knock-in rat model for glutaric aciduria type I allows further insights into pathophysiology in brain and periphery

17:15 Closing Remarks – followed by Apéro dînatoire

*SGIEM Meeting sponsored by:
(in alphabetic order)*

