



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) – **ONLY ONLINE POSSIBLE**

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

Tel: 031/632 52 45

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 – Members only)

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; Credits on registration only)

Scientific Symposium

Venue: **ONLY 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 – unfortunately not followed by Apéro dînatoire

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

