

General Meeting of SGIEM, Thursday 27th August 2020

Morning

Annual meeting of the SGIEM National Network

Venue: Auditorium Murice E. Müller (MEM), Murtenstrasse 35, Entrance 42

09:15 General Meeting, Part 1

10:15 Coffee Break

10:45 General Meeting, Part 2

11:45 Lunch

Subgroup Meetings 12:15-14:00

Conference Room Vista:	12:15-14:00	Dietician Group
MEM Auditorium:	12:15-13:00	Adult Group
	13:00-14:00	Mitochondria Group
Aquarium: (Floor E, Kinderklinik)	12:15-14:00	SGBOND

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Afternoon

Scientific Symposium

Venue: Auditorium Murice E. Müller, Murtenstrasse 35, Entrance 42

14:00 **Opening** of the Scientific Symposium, Matthias Gautschi

14:05 **Prof. Hans Jung, Zürich (by WebEx; time slot may change!):**
Mitochondrial Disorders in Adult Neurology

14:45 **Maria Isis Atallah Gonzalez, Lausanne:**
Spinal cerebrotendinous xanthomatosis: an easily overlooked treatable disorder

15:00 **Florin Bösch, Zürich:**
Health-related quality of life in paediatric patients with intoxication type errors of metabolism: Analyses of an international dataset

15:15 **Timothée Ndarugendamwo, Zürich:**
Understanding the clinical spectrum of Osteogenesis imperfecta,

15:30 Break

16:00 **Tatiana Brémova-Ertl, Bern:**
Heterozygous mutations in lysosomal storage diseases and their role in late-onset neurodegeneration and neuroinflammation.

16:30 **Pei Jin Lim, Zürich:**
Defining Pathomechanisms and Molecular Signatures of X-linked MBTPS2 Osteogenesis Imperfecta

16:45 **Jan Gaugler, Bern:**
The impact of genotype on the deep phenotyping of the Swiss Classical Galactosemia Cohort

17:00 Closing Remarks – followed by Apéro dînatoire