General Meeting of SGIEM, Thursday 30th March 2023

Morning

Annual meeting of the SGIEM National Network (for Members only)

Venue: University of Bern, Hallerstrasse 6, 2nd Floor, in Bern

Agenda : see separate mailing to all members

09.15 General Meeting, Part 1

10:15 Coffee Break

10:45 General Meeting, Part 2

11:45 Lunch in Foyer

Subgroup Meetings 12:00-13:00

Seminarraum 203:	12:00-13:00	SGBOND
Hörraum 205 (Entrance B):	12:00-13:00	Adult Group
Alpenraum in WTI (GF):	12:00-13:00	Dietician Group
Hörraum 205 (Entrance A):	12:00-13:00	Labs
Foyer:	12:00-13:00	PO

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Afternoon

Scientific Symposium (open to all interested; on registration only)

Venue: University of Bern, Hallerstrasse 6, 2nd Floor, in Bern

13:15 **KEYNOTE LECTURE:** Chair: Christel Tran

Beyond the bones: The 2023 nosology of genetic skeletal disorders and its relationship to metabolism and genetics

Prof. Dr. Andrea Superti-Furga, Lausanne

14:15-15:05 **SESSION 1: Diagnostic and Clinical Practice** – Chair: Marco Strasser

14:15 *Christian Urzì*: Intra- and Extracellular Metabolic Investigation of Perfused Fibroblasts by Real-time NMR Allows for Better Discrimination of Mitochondrial Defects

14:30 *Aurel T. Tankeu*: Biotinidase Deficiency: What Have We Learned in 40 Years?

14:45 *Christina Kaufman*: Diagnostic delay in patients with cerebral creatine deficiency disorders

15:05-15:35 Break

15:35-16:20 **SESSION 2: SGBOND & Porphyrias–** Chair: Aurel Tankeu

15:35 *Manuela Messikommer*: Late diagnosis of childhood-onset hypophosphatasia in an adult with recurrent fractures: the impact of enzyme replacement therapy

15:50 *Giulio Marcionelli*: Perturbations in fatty acid metabolism and collagen production infer pathogenicity of a novel MBTPS2 variant in Osteogenesis imperfecta

16:05 *Jasmin Barman-Aksözen*: Quality Adjusted Life Years in erythropoietic protoporphyria and other rare diseases. A patient-initiated EQ-5D feasibility study.

16:20-17:05 **SESSION 3: Potential new treatments** – Chair: Giulio Marcionelli

16:20 *Alexander Laemmle*: Using induced pluripotent stem cell (iPSC) technology to understand the ultra-rare metabolic disease malate dehydrogenase 2 (MDH2) deficiency (MDH2D) and its potential treatment with triheptanoin

16:35 *Gabriella Fernandes-Pires:* Rescue of myocytes and locomotor activity through intracisternal AAV9 gene therapy in a rat model of creatine transporter deficiency

16:50 *Marco Strasser*: Individual therapeutic trial for a severe sleep disorder in a patient with mitochondrial disease using a rocking bed

17:05 **Prize best presentation** / Closing Remarks – followed by **Apéro dînatoire**