

Minutes of the

SGIEM General Meeting 5th July 2018, 10:15-12:30

Location: Inselspital Bern, Wirtschaftsgebäude, Floor H, Room Paradiso

Present: Diana Ballhausen (DB, Secretary), Barman-Aksözen Jasmin, Matthias Baumgartner (MB), Bilz Stefan, Bosshard Tina, Burda Patricie, Christ Emanuel, Felser Andrea (new member), Fernandez Susana, García-Segarra Nuría, Matthias Gautschi (MG, President), Giunta Cecilia, Graf Stephanie, Hochuli Michel, Hundsberger Thomas, Ilse Kern (IK), Lämmle Alexander, Link Bianca, Deborah Mathis, Minder Elisabeth, Moor Daniela, Nowak Albina, Jean-Marc Nuoffer (JMN, Treasurer), Marianne Rohrbach, Salvisberg Claudia, Serratrice Christine, Christel Tran (CT)

Apologies for absence: Luisa Bonafé, Ralph Fingerhut, Forny Patrick, Johannes Häberle, Oswald Hasselmann, Jaya Heinzmann, Martin Hersberger, Kurth Sandra, Fabian Meienberg, Robert Steinfeld, Gabor Szinnai, Beat Thöny, Nina Zeltner

Absent: Bärlocher Kurt, Barbey Frédéric, Bosanska Lenka, Braissant Olivier, Chambru Charlotte, Cremonesi Alessio, Fowler Brian, Huemer Martina, Jung Hans, Krayenbühl Pierre A., Lenherr Nina, Palla Antonella, Perret-Hoigné Eveline, Refardt Julie, Rösler Kai, Rymen Daisy, Sluka Susanna, Superti-Furga Andrea

1. **Welcome, additions to agenda (MG):**

2. **Minutes of previous meeting (MG):** accepted, no comment.

3. **Matters arising from previous meeting (MG):** none.

4. **Statutes (MG):** A German version of the statutes was sent out before the meeting. The French version was missing. Minor changes are proposed to be implemented into the statutes under Artikel 10: "Vorstand...inclusive dem Präsidenten und dem Schriftführer (Secretary)." The proposition is accepted with 24 voices and 1 abstention, no oppositions. The new version will be circulated by email in German and French.

5. **Revisors (MG):** Michel Hochuli and Alexander Lämmle are proposed as revisors for the budget of the SGIEM. 22 persons agreed, 3 abstentions, no oppositions. Both, Michel Hochuli and Alexander Lämmle accepted to be revisors.

6. **Finances 2017 / Budget 2018 (JMN):** The final balance on 31.12.2017 was CHF 71'285.-. The participants accepted the balance 2017 unanimously as far as the revisors confirm that all is fine. The budget 2018 is highly discussed. A sponsoring of CHF 50'000.- per year is needed to cover the costs for the biannual meeting ASMM (about CHF 50'000.- every second year) and the yearly running costs for the secretarial office, the web page and the register. So far Actelion, Alexion, Biomarin, Danone, Sanofi, Shire and Sobi are sponsors of the SGIEM.

7. **Revisors report:** the new revisors will report to the steering committee if all is fine.

8. **Secretary's Report / Update on membership (DB):** The SGIEM counts currently 58 members. The members' list and contact details will be available on the SGIEM website with access limited to SGIEM members. The following persons were accepted to become new members of the SGIEM:

Nina Lenherr, Basel

Jaya Heinzmann, Zürich
Christine Serratrice, Geneva
Robert Steinfeld, Zürich
Nina Zeltner, Zürich
Stefan Bilz, St. Gallen

9. Work groups:

PKU guideline group (DB): Publication of a DACH guideline in the final phase. Any interested SGIEM member is invited to participate. Michel Hochuli left the group and does not want to join it again.

kosek (MG): Inborn errors of metabolism and neuromuscular diseases are the two pilot projects of the kosek (=Nationale Koordination Seltene Krankheiten) to elaborate the structure of reference centers and networks.

Rare Disease Registry (MB): So far, a grant to create the infrastructure has been obtained. Further applications for money are currently submitted as bridging financing is needed for 2018/2019. MB is optimistic that the registry will be realized but it is not secured yet.

SGIEM Website (IK): Not online yet. An email has been sent out to the members asking them to provide the necessary information. Not all have replied yet. The global structure of the website will exist in 4 languages (German, French, Italian and English). The website is intended to address to patients and health professionals. An external professional person is needed for critical review. It is important for the sponsors that this website will soon be online. It was decided that the emails will be sent out again with clear instructions what to do.

Mitochondrial disease guideline (JMN): not yet published. JMN will give a proposition on how to handle it for Switzerland and will circulate it.

KSME (DB, Daniela Moor): Last version of June 2018. No letter of support needed from the SGIEM for new products*. An update will only be done once a year, always in January. Mrs Rajower from the BSV asked the SGIEM to communicate which products on the KSME are not on the market anymore. The dietitians will prepare an email addressed to Mrs. Rajower by the end of 2018 indicating all obsolete products on the KSME.

* **Added in proof:** apparently, a letter of support is nevertheless needed, on unclear grounds; and was delivered by us on behalf of the SGIEM for Ceres MCT Oil (Dr. Schär).

Lab list (Deborah Mathis): the application was done last year, two rounds of questions have already been answered. There is hope that the recommendation will be accepted in October 2018 by the BAG, so that it could be on the list in 2019. The recommendation describes different positions depending on the technique used to analyze. It includes the possibility of subtreating analyses that then could be sent abroad. The Kinderspital is currently setting up the analyses of S-adenosylmethionin and S-adenosylhomocystein and currently accepts to analyze samples free of charge.

Newborn screening (MB for Ralph Fingerhut):

CF: a pilot project was performed in the last 5 years (2013-2018). It is now on it's way to the Analysenliste.

SCID: The demand to screen for SCID is waiting for final approval for reimbursement and might be ready to start on the 01.01.2019.

Biotinidase deficiency: the screening method changed 5 years ago. The new assay is more sensitive and more patients are tested positive. This is due to a frequent mutation that leads to partial deficiency. The current recommendation is to treat patients with residual activity <30%, to consider patients with residual activity <10% as a complete deficiency and to not treat patients with a residual enzyme activity of > 30%. The question of overtreatment is difficult to answer. Following reports from Prof. Barry Wolf (USA) some patients with partial deficiency developed severe neurological symptoms. A cut-off of 20% is used in the Netherlands. There is a certain maturation of the enzymatic function during the first weeks of life. A second control at the age of 3-4 months seems to be indicated in patients with partial deficiency. The result is influenced by pre-analytical treatment. Patricia Burda highlighted the importance of centrifugation, and freezing of the blood sample before shipment on dry ice (see protocol attached).

10. Sub-groups :

SALS (Marianne Rohrbach):

Mr Wiesbauer asked whether the SALS would agree to help with the organization of the international MPS Congress to be held in Switzerland in 2022. An average of 1'000 participants come to this congress. The SGIEM agreed to provide scientific advice.

Mrs Sattelberger from Shire wants lyso-Gb1 to become available as a diagnostic parameter in Switzerland. No development is so far foreseen in Switzerland and with the expected modification of the Analysenliste it might become possible to send it abroad.

Mr Coppex asked the SGIEM to provide help with the registration procedure for ERT for alpha-mannosidose. There is one adult patient in Bern, one transplanted patient in Lausanne and some more in Zürich. The SGIEM agreed to help with the registration process in Switzerland. Mr Coppex should contact Marianne Rohrbach and Diana Ballhausen to discuss the needed action in more detail.

Adult group (CT): had their meeting in the afternoon after the general meeting

Dietitian's group (Claudia Salvisberg): also had their meeting in the afternoon. They presented a consensus on the diet for adolescent (>14 years) and adult galactosemia patients that was unanimously accepted by the SGIEM.

OI group: no news, no representative present at this meeting

Others: no.

11. Next meeting : March 2019 in Bern

Diana Ballhausen, Secretary
For the Steering Committee

Supplement after this meeting: the revisors Michel Hochuli and Alexander Lämmle have reviewed the financial statement 2017 and confirmed their approval by signing the statement in October 2018.