



MINUTES
STRATEGIC ADVISORY GROUP MEETINGS | JAN/FEB 2022

VIRTUAL | FEBRUARY 15, 2022
19.00 – 20.00 CET

RARE DISEASES

Chaired by Risto Lapatto & Liesbeth Siderius

Altogether 20 people attended part or all of the meeting, list of registrations attached

Presentation of Risto Lapatto [can be viewed here](#).

Presentation of Liesbeth Siderius [can be viewed here](#).

1. Participants were welcomed, and agenda was approved.
2. Minutes of the spring meeting 2021 were approved
3. Tim Buckinx is a Belgian entrepreneur and foremost father of a son with refractory Ring 20 (Orpha: 1444) epilepsy gave a presentation with a title: “Dad, can you create a light that turns on when my brain switches off?”
“Dad, you work in digital, can you create a light that turns on when my brain switches off? Because then my teacher isn’t angry at me anymore.” The biggest impact on his daily life was people not noticing his absence seizures. The digital therapeutics company epihunter (www.epihunter.com) combines wearable EEG with AI in a smartphone app to detect and signal absence seizures in real-time and collect the EEG data and automated seizure videos (recorded at home) in a portal for the physician. The platform has been used by more than 400 end-users and is available in Europe and Australia/New Zealand. A recent multi-centre prospective clinical validation study will be published next month in Epilepsia, showing a median sensitivity of 93% and a median false alarm rate of 0/h. Reach out to tim.buckinx@epihunter.com.
4. UEMS Multidisciplinary joint committee on rare and undiagnosed diseases. Risto Lapatto is representing the EAP and reported on the recent activities of the MJC. MJC has become more active during 2021, and it is developing its own training programme with a test. Furthermore, some members are actively pursuing a European Board of rare and undiagnosed diseases. There are also projects on some rare diseases lead by committed individuals, and a memory of understanding is being developed between UEMS and EURODIS representing patients. The UEMS will take decisions on MJC, but the EAP will watch the developments and make sure that children are duly remembered.
5. News from the European Reference Network’s (ERN’s) by Risto Lapatto. The long application process of new healthcare providers joining the 24 ERNs is finally over, and all ERNs have grown substantially. The first five years have been used to establish the ERNs, and the second term, with new members and a new grant from the EU, should yield more results. ERNs are divided into Work packages such as coordination, training, research, patient involvement etc, and Thematic areas that depend on the ERN. Success depends on voluntary work by professionals. There are issues with data transfer, but the ERN system is



so large, that the EU is actively solving problems. Countries outside EU can collaborate with ERNs.

6. ERN Expert platform on newborn screening: Risto took part in a meeting Towards European Recommended Uniform Screening Panel (ERUSP). The goal of this group, that works together with International Society for Newborn Screening, is to develop definitions of cases, create registries and make recommendations on screening panels. There is a wide distribution of panels in use in Europe, even though most people agree on ethical issues. Genetic screening is coming, and that has to be carefully dealt with.
7. Proceedings on digital child health : the HL 7 community and International Patient Summary, by Liesbeth Siderius. World Health organization recommends on home-based records for maternal, newborn and child health. HL7/ FHIR is a digital standard to exchange data with international classifications. The International Patient Summary (IPS) of HL7/ FHIR is an electronic health record extract, containing essential healthcare information about a subject of care. The IPS is building bridges between “ home “ health and care environment and any other place where the patient needs to visit a clinical professional whether within or across borders. A design of a rare disease IPS is discussed. Presentation attached.
8. Other Business and General Discussion: Important is patient/ family involvement. Transition from child to adult care is an issue in Poland. WHO might get involved considering the macro-level of rare diseases, the home based record and the data exchange. In Russia there are networks on rare diseases as well. It is suggested to advocate for a digital rare disease card, to ensure each person with a rare condition gets the care needed, like in Portugal. A platform with information for families/patients and physicians would be helpful.
9. The group will hopefully meet in person in May during the EAP Spring meeting.



List of Registered Participants:

First Name	Last Name	Registration Time
Jernej	Završnik	2/15/2022 13:01
Olga	Cirstea-Buga	2/14/2022 17:35
Adib	Salim	2/13/2022 12:55
Marina	Mamenko	2/15/2022 9:40
Nataliia	Samonenko	1/20/2022 12:22
Larisa	Prikhodina	2/15/2022 12:35
Stefano	DEL TORSO	2/15/2022 19:02
Miguel	Martins	2/14/2022 16:17
Suja	Somanadhan	1/19/2022 12:58
Tim	Buckinx	1/11/2022 20:37
Liesbeth	Siderius	2/15/2022 18:45
Maria	Gutu	2/15/2022 0:23
Jolanta	Wierzba	2/5/2022 9:06
Urh	Groselj	2/14/2022 18:52
Risto	Lapatto	1/24/2022 19:00
Jordi	Antón	1/20/2022 10:20
Violeta	Iotova	2/15/2022 18:57
Ivana	Kavecán	2/15/2022 10:24
Adamos	Hadjipanayis	2/14/2022 19:02
Artur	Mazur	1/22/2022 12:30
Joe	Brierley	2/15/2022 15:54
Manuel	Katz	2/15/2022 13:58
tommaso	ballerini	2/15/2022 19:51
Sergey	Sargsyan	2/15/2022 7:58
Ann	DE Guchtenaere	2/15/2022 19:02