European Academy of Paediatrics
Paediatric Section of U.E.M.S (European Union of Medical Specialists)

Minutes
EAP Winter Minutes- Rare Diseases
Friday, 7 December, 2018 | 11.30-12.15

1. Agenda, approved

7 December Patient Solidarity Day in 2018 the theme is " *Safe Medication and Healthcare for All ‘* .

All attendants are ask to raise hand to support the Patients and their representatives

2. It can be done differently!

Kees van der Graaf father of a son with FSHD, a progressive muscular disease with no therapy presented. Since 2006 he devoted his time, his experience as president of a multi-national and his worldwide network to start the research to establish a therapy for FSHD: ‘a pill’ . In 2013 he established the bio-tech start-up Facio Therapies (www.facio-therapies.com). The board of this company consists of wealthy patients with FSHD and/or their family members. Instead of the usual business pharmaceutic business model the goal is develop an affordable medicine (“the lowest possible price”) and future profits will be used to fund quality of life enhancing projects.

3. Follow up om Spring meeting Sofia, Bulgary 2018
- **Access to health** document has been prepared and has to be converted as an article as well as letter to the EU.
- Pediatricians in have been in Rare Diseases Macedonia, Slovenia, Sri Lanka, Germany, The Netherlands, Poland (report has been submitted for the EAP new)s.

4. UEMS **MJC Rare and Undiagnosed Diseases** (RUD) Report
Since October 2016, the first meeting of the MJC RUD, the EAP is represented as Specialist Society. At the moment there are two pediatricians representing a national medical association (NMA) in the MJC RUD.
Summer 2018, there has been a survey among the members of the MJC RUD on training requirements regarding Rare Diseases (the results are attached to the minutes). 14/ out of 34 responded. All agreed that a multidisciplinary approach should be included in a standardized core training on rare diseases.

At the 2018 meeting it was proposed to develop an ETR Rare Diseases. Documents to support this suggestion are a lacking at this moment.

The CME-CPD in BRUSSELS on 23-24 November as part of the 60 th was attended. Many new concepts and collaborating partners where presented. Among them patient involvement.

5. Other business

E Health : The UEMS has established (or refreshed) an E Health working group. This working group has been attended. Harmonization and interoperability of data is an important issue in the field of rare diseases.

As was discussed with Peter Alorjay during the MOCHA project, HL7 is a tool to support transmission of data such the patient summary.

Discussion:
The EU has just announced to (large) budget for the rare diseases. Access to diagnostic tools should be available. Pediatricians often are the first contact of child with a rare condition. Down syndrome Rett syndrome and many other disorders are diagnosed in pediatrics. When new techniques become available the diagnostic process should be clear and efficient.

The EU is at the moment asking feedback on **E-Health recommendations**. How electronic health records can be accessed and shared more easily in every EU member country. To make it easier for patients, health professionals or other authorized parties to use health data from different records across the EU – for example, to avoid having to repeat medical tests that have already been carried out in one country. Give FEEDBACK until 20 December 2018.

The EAP should speak for those children born with a pre-existing condition, especially in times where economics in health care are the driving force. The disabled children will be easily neglected.

December 21, 2018