

Minutes Rare Disease WG (EAP, UEMS Section of Paediatrics)
Part of the rare disease/ ethic WG session
Brussels, Friday December 1st 2017, UEMS building

1. Attendance
2. Minutes Spring Meeting Vilamoura, Portugal May 12th, 2017, no comments
3. Report UEMS Multidisciplinary Joint Committee Rare and Undiagnosed Diseases

Mission

The aim of the MJC 'Rare and Undiagnosed Diseases' is to promote high quality multidisciplinary health care for European citizens affected with rare or undiagnosed conditions.

Objectives

To **improve the awareness of the multidisciplinary** nature of rare and or undiagnosed conditions among national societies, organisations responsible for training and CME and the European Commission.

To **raise standards of specialist training** in rare or undiagnosed conditions by:

- establishing standards of training
- ensuring these standards are met by processes of accreditation, documentation and visitation
- ensuring that trainees have a comprehensive training according to UEMS/EBR

To **maintain standards of clinical care** by recommendations for requirements for CME

To ensure provision of appropriate and **high quality educational** activities, as well as setting and monitoring standards for courses, meetings and other educational activities. Clinical Genetics; Internal Medicine; Neurology; **Paediatrics**; Paediatric Surgery; Pharmacology; Psychiatry; Rheumatology; Oral Maxillary Facial Surgery; Ophthalmology (33 section and country representatives).

The right to access to highest standards of care of the most vulnerable children:

4. Making Roma Health and Early Childhood Development a priority for Europe.

Marius Lucian Tudor

Roma Health and Early Childhood Development Project Manager

European Public Health Alliance (EPHA)

50% of the Roma children do not receive the mandatory vaccinations. 30 % of the pregnant woman do not have medical checks. 30% of the children are not visited for pediatric care. The Roman representatives are member of the European Public Health Alliance. Through this organization there we a collaboration with the EAP (also member of EPHA). A personal story illustrates how Roma's do not profit from new diagnostics and treatments. Still Roma children have the same rights of access to adequate health. Better training in pediatrics in areas where the Romas are living would help. Pediatricians should know about the Roma culture.

Besides there is a lack of vaccines. Discussion : this could be due to the fact that vaccines are sold at the black market in some countries.

Children with disabilities should also be able to profit from special school. Because of the ethnic origin and language differences they are often left out.

5. From first feature, to gene, to society

Introduction Info Web Rare Diseases

Petra Poulissen, Patient Representative

The Netherlands

Petra presents her life story. Being born with some congenital anomalies, not hearing well at primary school, restricted in spinal movements. Finally in her twenties she found her diagnosis by own research.

All these different features together build up to the diagnosis Goldenhar Syndrome.

More information on diagnosis and care coordination would help the patients and families with a rare condition.

6. Orphan drugs and pediatric regulation (extra)

Lenneke Schrier explained how LOW, the agency working for the EAP, informed the EAP on reports on pediatric medicine. LOW had notified the EAP there would be an important European Ministers meeting December 9th.

A statement and draft paper on orphan and pediatric drug regulation was prepared. A three point statement was adopted to present at the GA. Still open for suggestion to improve.

Present:

Christine Aebi	Switzerland
Ivan Bambir	Croatia
Ulrike Gaiser	Germany
Risto Lapatto	Finland
Jan Lebl	Czech Republic
David Neubauer	Slovenia
Pierre- Andre Michaud	Switzerland
Jelica Predosevic	
Kadre Saare	Estonia
Lenneke Schrier	the Netherlands
Liesbeth Siderius	the Netherlands
Tom Stiris	Norway
Arunas Valiulis	Lithuania
Norbert Varga	Hungary
Viri Zeman	Czech Republic

Guests:

Petra Poulissen	the Netherlands
Klaus Rose	
Marius Tudor	Belgium/ Roma

LS December 16th, 2017