



## MINUTES

### EAP Winter Meeting 2019 | Brussels, Belgium

#### Rare Diseases Working Group

Friday, 6 Dec 2019 | 12.30 – 13.30

Chairs of Rare Diseases Working Group:  
**Risto Lapatto & Liesbeth Siderius**

1. Welcome and Approval of Current Agenda & Minutes of the Spring Meeting in Vilnius, May 2019  
Agenda was approved as were minutes of the working group's Spring meeting 2019.

2. Update on European Reference Networks

Risto Lapatto reminded audience about European Reference Networks and explained their role. The participants were encouraged to find out about ERN members in their own countries.

3. The Impact of a Diagnosis: A Personal Account

Saskia de Koster and her daughter Joojje Kist from the Netherlands gave a touching story on the life with their son/brother who got a molecular diagnosis of his disease more than 20 years after symptoms presented. The importance of diagnosis was clearly presented. The support and information from the facebook patient group has been very important.



4. Sustainable Development Goals, Universal Health

Coverage and Rare Diseases: What is the Least Paediatricians Can Contribute?

Liesbeth Siderius introduced the Universal Health Coverage (UHC) as adopted by the United Nations in September 2019. She then showed the results of the survey, done on the attitudes of EAP members on key UHC issues. 12th of December will be the UHC Day.

5. General Discussion

Audience participated in a lively discussion on importance of genetic diagnosis, use of Big Data, artificial intelligence and interoperability with international classifications.

Presentations will be loaded on the EAP Rare Disease web page.