Brussels, Friday January 29th 2016,
WG Rare Diseases 11.00- 13.00 **HALL B**
The Royal Windsor Hotel, Brussels, Belgium

1. Attendance and Apologies: no apologies ; agenda accepted
   The meeting is dedicated to

> UNICEF PLEA, November 2015, *In 2015, an estimated 5.9 million children will have died before reaching age 5. Children from the poorest families are, on average, nearly twice as likely to die before their fifth birthday as those from the wealthiest families. Children born in rural areas and those born to mothers with no education are more likely to die before reaching age 5 than those born in urban areas or to educated mothers.* **Improving data collection and analysis** *Disaggregated data – that is, data broken down by subcategories rather than whole populations – are at the core of efforts to realize the equity agenda.* The **European Academy of Paediatrics considers sustainable strategies to be essential to promoting early childhood health and development**. It wishes to contribute to sustainable strategies that build on multisectoral approaches for guaranteeing the rights of children with disabilities and their families. These strategies should include: identifying children with disabilities through primary care, child health surveillance, and vaccination programs;providing children with an early and accurate diagnosis of disease; and developing management programs to prevent disabilities and to achieve the maximal potential of the child throughout its life. There is the urgent need to provide professionals and families with guidelines of diagnosis, medical and social care through an quality guaranteed open resource **to ensure the same level of access, harmonization and standardization.**

2. Minutes Meeting Bratislava, 2015: no remarks

3. The Importance of Open health data platform for developing comparable interoperable patients registries rare diseases
   Anze Droljic, MARAND, Slovenia http://www.marand-think.com/
   Presentation https://www.dropbox.com/s/3cof83q51st7cuj/think_registry_2016.ppt?dl=0
   Comment: from Metaboloc Disease UK (Morris) : three different metabolic registries: held by parents, drug companies and (expensive) European funded network (homocystinurea)
   The RD group and Slovenian participants will prepare next steps on harmonization for the next meeting in Dublin.

4. Report from the EU Expert Group on Rare Diseases (12-13 November 2015)
   CROSS BORDER GENETIC TESTING OF RARE DISEASES IN THE EUROPEAN UNION established:
   -Obtaining an accurate and timely diagnosis is a priority for all people with a potentially genetic RD; therefore, access to genetic testing -whether provided locally or on a cross-border basis - should be ensured, to facilitate such diagnoses, when there is a clear clinical indication.
   -The expert group underlines the importance of assessing genetic testing, on the basis that early diagnosis through clinically-guided genetic testing may avoid the need for further invasive and/or unnecessary exploratory and therapeutic procedures
   -Whether genetic testing is provided on the national/regional level or on a cross-border basis, expertise should be shared at the EU (or global) level.
   -Appropriate information on genetic testing laboratories should be made available to facilitate crossborder genetic testing of rare diseases, particularly when pertaining to the quality of laboratories.

5. What are your experiences & expectations of Expert Centres and Reference Networks?
   http://ec.europa.eu/health/ern/events/ev_20151008_en.htm
   http://ec.europa.eu/health/ern/docs/faq_establishing_ern_en.pdf
   - Lincke, Holland : In the Netherlands expert centres are designated , now there is a problem of the grouping of one rare disease in different categories
   - Zeman, Czech Repulic : the are many inborn errors of metabolic diseases; the more high economy countries will have enough experience for all rare diaseses
   -Dzivite, Latvia : lower economy countries may have difficulties to meet the criteria for the European reference centres

6. Rare Best Practices is making progress there will be a repository on rare disease guidelines appraised with the AGREE method
   http://www.rarebestpractices.eu/
   Please note that project end this year.

7. Importance of Primary Care Pediatricians for children with a rare and disabling condition and early development
   Making Patient- Centered Community Care a Reality
   Melina Raso, Health First Europe
   Discussion : EAP joining Health First Europe? What could HFE mean for EAP?
   -Lincke, Holland: does not see any good in budget spending organisatie
   -Raso: the membership is for free, members are just asked to put their knowledge into a the common cause
   -Aebi: could we collaborate on the refugee's emerging in Europe? Yes
   - other subjects mentioned : vaccination, antmicrobial (mis) use
   - Ivanov: the issue of community based genetic testing – a cost effective analysis – related to the knowledge of rare diseases
   European Disability policy
   Inmaculada Placencia Porrero, DG EMPL
   Presentation on the new sights of the universal disability act.

Ivan Ivanov	Bulgary
Jiri Zeman	Czech Republic
Peter Hoyer	Germany
Peter Oldrich	Germany / Spain
Iveta Dzivite-Krisane	Letvia
Carsten Lincke	Netherlands
Liesbeth Siderius	Netherlands
Dana Crain	Romenia
David Neubauer	Slovenia
Jernej Zavrsnik	Slovenia
Tadej Avcin	Slovenia
Isabel Leiva	Spain
Christine Aebi-Ochsner	Switzerland
Andrew Morris	UK
Leonid Dubey	Ukraine
Mohammed Rashid Salem Elnaddoury	UAE

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