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

Rare Disease Working Group (EAP, UEMS Section of Paediatrics)
Friday May 30th 2014 8.15- 9.45 Rzeszow, Poland

Minutes (draft)

1. Attendance and appologies: no appologies, agenda was approved

2. Minutes Meeting Brussels December 2013: no comments

3. Update on RARE-Bestpractices (www.rarebestpractices.eu):
   Michele Hilton Boon, Healthcare Improvement Scotland, Glasgow, Scotland
   "Searching for rare disease guidelines: new developments from RARE-Bestpractices".
   Presentation is included.
   During the presentation quetions where posed:
   Where do you currently go if you want to find a guideline on a rare disease?
   Orphanet for acute care; consult the expert; Google; patient organisations
   How do you think you might use a database of research recommendations?
   What would it have to offer to be useful to you?
   Quality; child and patient registries
   As users of the guidelines database, how valuable would information on guideline quality be to you? What do you see as the most important issues in the evaluation of rare disease guidelines?
   Some discussion on the possibility to develop guidelines on diseases where there is limited evidence: methods as are AGREE are questioned. According to the Rare Best Practice group it is possible based on expert opinions including patient experience.

4. Update on European Commission Experts on Rare Diseases;
   The EAP considers sustainable strategies:
   identifying children with disabilities through primary care, child health surveillance
   providing children with an early and accurate diagnosis of disease; and
   developing management programs to prevent disabilities
   David Neubauer: (ethics group)
   How can electronic records help; possible collaboration with PARENT project?
   At present the ethics group has no presentation on the electronic records.
   David (and Liesbeth) has been in contact with Matic Meglic, pubic health, Slovenia to collaborate on harmonization of data collection for European registries on behalf of EAP. The group present advises to proceed on the issue of harmonization and standardization.
   Suggestion is to invite Matic Meglic at the next EAP meeting in Brussels.

5. Any other bussiness and proceedings
   Each individual present is asked what the status of the national rare disease program is:
   - Czech republic: the (large) primary pediatric group colleborates with metabolic and genetic specialist, the National government is supportive.
   - Spain : patients with rare diseases are registred by their physian, once registred they get care funded. FEDER is the governmental organisation that registers. One of the problems is the early diagnosis. Neonatal sceening programms differ per region.
   - Latvia: One hospital; there is no collaboration between the care providers

- Ireland: only hospital care , no primary care peadiatricians; pediatricians are not involved in a national RD program; the availability of guidelines would be welcome
- Slovakia: integrated care between primary care and centres; similar to Czech republic
- Slovenia: rare disease program with centres: CF , Fabry at University Hospital; no national registry; registration at level of univ hospital
- Israel: large group of primarycare paedistricians (900) the work in close contact and consult eachother as wel with hospital based specialists, for information Google is used
- Austria: 8 milj people, children with rare disease are taken care for by subspecialists : for example 3,5 CF centres; they all register in a different way, when a child moves to another region it will be in a different registry
- Netherlands: at present there are difficulties in assigning the national centres of expertise

It is concluded that in the different countries there are still need to:
- improve early diagnosis/ identification of the child with rare diseaes in primary care
- improve care coorination
- get epidemiological data
- harmonize and standardize data collection.

The last point should have to priority. Every one is ask to contribute the a new rare disease project as folow up on the one we are involved.

6. Next meetings
   October 16-17: Invitation Michelle Hilton – Boon: Edinburgh Scotland : meeting Rare Best Practice guideline searches RBP WP 4
   October 18 EAPS Barcelona : Workshop Rare Diseases
   Devember 5-6: Brussel EAP

Liesbeth Siderius
June 2014

Attendance:

Michelle Hilton- Boon	Scotland (Healthcare Improvement Scotland, Glasgow)
Manuel Katz	Israel
László Kovács	Slovakia
Gabriella Kubatova	Czech Republic
Isabel Leiva	Spain
Carsten Lincke	Netherlands
Vizma Meisane	Latvia
David Neubauer	Slovenia
Ane Nevos	Portugal
Liesbeth Siderius	Netherlands
Martin J White	Ireland
Max Zach	Austria