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

Minutes Rare Disease WG
(EAP, UEMS Section of Paediatrics)
Brussels, Friday December 2rd 2016,
WG Rare Diseases 11.00 - 12.30 **HALL B**
Hilton Brussels Grand Place, Brussels, Belgium

1 Attendance and apologies

2. Minutes Spring Meeting Dublin; no remarks

3. ICF-CY as a problem solving tool in complex pediatric cases
Janneke Brandt, project assistant of ICF-CY Meduse project (www.icfcy-Meduse.eu), Medical School Hamburg, Germany
The internet-based tool build in the Erasmus + project www.icfcy-Meduse.eu is free available in English, German, Macedonian, French, Slovac, Italian, Portuguese and Turkish. Further language variants can be easily integrated if ICF-CY excel-files are available. The tool is for a multidisciplinary use, it associates anonymous narratives of parents with ICF-CY codes (by means of dropdown menus, full text search functions and selection of code-sets) and allows an evaluation/graphic representation of the situation of a child with a health problem in his/her relevant environment. Anonymized data can be individually integrated as pdf-files into the local individual patient-documentation. To use the tool a 2 days training is necessary – including an introduction into the general philosophy and ethical guidelines of ICF-CY. During the session a case was demonstrated in an interactive manner. Pediatricians seem to be more focused on the child and may not notice the environment. The ICF-CY might be of used to define support/treatment needs of children with disability. Parents are understood as a part of the Team around the child and in German and Macedonian parent-friendly versions of the tool are available. The tool uses WHO-qualifiers (.0 till .4) in terms of the quantity/quality of the health problem, facilitators are scored from 0 to +4. The ICF is used in Armenia, Switzerland and in the Netherlands by physiotherapists and speachtherapists. For further information: Manfred.pretis@medicalschool-hamburg.de (responsible) or janneke.brandt@medicalschool-hamburg.de (project assistance)

4. Report from the EU Commission Expert Group on Rare Disease
(November 28-29th 2016, Luxembourg)

Change of **regulation on the EU expert groups**: www.cc.cec/regexp/welcome.do . Any organization needs to be registered. EAP can be registered and delegate representatives to the (different) commissions. http://ec.europa.eu/transparency/regexpert/. About the continuity after the end of 2016 of the RD expert commission is not decided.

Patient organization (EURORDIS) did an **access campaign**: it seems the RD strategies have so far not has the positive effects one would have expected.
HTA discussion is dominated by **orphan drugs** and the differences in reimbursement procedures in each country.

Report of member states: each member state has developed its own criteria on the basis of EUCERD criteria. It remains unclear whether these criteria are the same and comparable. Still all countries have applied for ERN's. Member states have not seen each others applications procedures of the experts! The UK patient organization has a matrix ascertaining progress measuring the change. In europlan list of indicators are the most important way to measure the progress/ implementation.

Task Board of Member States
The Board of Member States, as laid out in the Commission Implementing decision, was set up on 5 February 2014.The main tasks of the Board are:

Approval of Networks proposals and healthcare provider's membership applications included in a Network proposal. Approval of healthcare providers wishing to join an existing Network; Termination of a Network (evaluation); Decision on loss of membership

Statement on behalf of EAP in the commission'meeting

Rare disease are common in pediatrics. Most children with a disability due to a chronic condition will have a rare disease. Most features of a rare disease will present in childhood to general primary or secondary care pediatricians. With the introduction of the European Reference Networks fragmentation of care lies in wait. What if the child has a complex epilepsy and a renal disease (tuberous sclerosis) or a hematological, gastrointestinal and skeletal manifestations of one rare disease?

All pediatricians are trained in normal growth and development as well as specialistic care such as nephrology , hematology, pulmonology, gastroenterology and endocrinology. It has been acknowledged that the care for the child is well of in the pediatric setting.

Pediatricians are sometime even obligated to care for persons far beyond the age of 19 years, because of the lack of sufficient trained generalist to take over coordination of care.

Within pediatrics there is a strong need for interoperable IT network to support the network the pediatricians already have developed within their society.

The disabled child needs to be vaccinated, growth and development should be under surveillance, common infections should be taken care for and participation in more than one ERN makes coordination by a generalist even more necessary.

With guidelines on diagnosis and care personal health pathways starting early in life are feasible since most IT tools are there. As the EU funded Rare Best practice project is showing elegantly rare disease guidelines exist. Over 200 rare disease guidelines where identified and assessed.

On behalf of the children with a rare and disabling condition I ask to take steps to support these children with up to date and open IT systems.

It is not a question of possibility, just an issue of priority.

News from EURORDIS: The United nations general assembly established : NGO committee for rare disease; at the right time; objectives of universal health coverage.

5. Report from the EU Commission Interoperability Workshop (November 30th 2016,Luxembourg)

Input in behalf of EAP:

All ERN's will need results of: laboratory test; imaging and pathology and other similarities as blood pressure and growth. Children may end up in different ERN's so these should be interoperable. Warning to assigns a so called UNKNOWN to a ERN, because it may prolong the diagnosis. Need to support patient centered records with different modules.

6. Report on Projects: FP7- Rare Best Practices : www.rarebestpractices.eu meeting

November 23- 24th Rome

EAP has participated in project that aims to

"Create a platform to improve the management of rare disease patients"

- Promote communication on rare diseases by disseminating trustworthy guidelines
- Identify rare diseases research needs
- Facilitate timely, effective and efficient translation of research into patient oriented clinical and public health practice

Over 200 guidelines on 43 different (groups of) diseases have been collected and appraised with the AGREE method. The guidelines and the appraisal can be found: http://www.rbpguidelines.eu/

In addition to the repository a model for guideline development for rare diseases has been tested with the GRADE method. A sicklecelldiseases guideline is produced in the pilot.

7. Report UEMS Multidisciplinary Joint Committee Rare and Undiagnosed Diseases

The kick-off meeting of the MJC RUD, 20th October 2016 Brussels.

The professional training on rare and undiagnosed diseases is presently weak. As high-quality education is one of the aims for the UEMS, the MJC-RUD could aim at enhancing training on rare and

undiagnosed diseases. The strength of the MJC-RUD is that it includes many specialties and can reinforce any produced documents on the European level through UEMS Council meetings.

8. General discussion activities in the field of rare diseases
- patient and family involvement - interoperability - how to proceed?

In Spain there is a great diversity of neonatal screening test differing per region.
The negative effects of fragmented care is been demonstrated with a case of a child with ambiguous genitals and bladder extasia. The child had been seen in 4 different academic hospitals, but no one has measured the child's length, at age 14 years the lengths was just 125 cm. Only the parents had kept a record of the child length over the years.
Rare disease programs in Switserland are developed under pressure of patient organizations.
In Portugal a Health electronic platform was created in the last few years allowing MDs and nurses to check the different consultations of the child in different settings. In the Pediatric Services, the Adolescent Consultation can centralize the follow up or eventually, the Private General Pediatrician in ambulatory setting there is one electronic health systems connecting hospitals and primary care.
In Armenia a recent case of atypical HUS demonstrated how physicians in collaboration with colleagues abroad successfully diagnose rare disease.
In Bosnia / Herzegovina there are RD activities coordinated at national level. The is a rare disease registry with now 160 patients registered. There is treatment for people with Pompe and Gaucher disease all in tertiary care.
In Ireland geneticist only do the diagnostic tests. They are not the ones who provide the care for the child. Transition from childhood to adulthood is still a challenge.
In Poland the ministry of health has established a national plan.
The university hospital in Slovenia is participating in 2 ERN's. The country has one electronic patient record shared in all hospitals but not with primary care. The HPO is included in the electronic record.
The situation in Lithouania is similar to Poland. Reference centres are assigned. In Hungary the non medical care is stronger developed than the medical.

In general there is a great diversity in what is actually done for the children with a rare and disabling condition. In theory it is possible to get insight in the care provided once care can be compared with help of interoperable health systems. The EAP could through its network provide insight in where in each country lie the possibilities and hurdles.

DRAFT December 30th 2016

Attendance

Christine Aebi- Ochsner	Switserland
Isabel Leiva	Spain
Andrea Luczay	Hungary
Artur Mazur	Poland
Pierre-Andre Michaud	Switserland
David Neubauer	Slovenia
Ana Neves	Portugal
Jelica Predojevic Sam	Bosnia and Herzegovina
Sergey Sargsyan	Armenia
Flemming Skovby	Denmark
Arunas Valiulis	Lithouania
Martin White	Ireland
Liesbeth Siderius	Netherlands