Provisional Agenda Rare Disease WG

(EAP, UEMS Section of Paediatrics)

Bratislava, Slovakia Friday May 29th 2015,
WG Rare Diseases 11.00- 12.30 **HALL A**

1. Attendance and apologies no comments

2. Minutes Meeting Brussels December 2014 (attached) no comment

3. Report from the EU Commission Expert Group on Rare Disease
Within the European Commission Expert Groep on Rare Diseases (CERGD) many issues are addressed that involve children thus we should or could address in EAP: Medicine for Children, Palliative care, E Health, social care. Understandable because chronic and disabling conditions are in rare in children, and most children with a disabling condition have rare and complex disorder. Report on issues special pediatric importance meeting March 12-13, 2015 :

A- Enrique Terol (DG SANTE D2) gave an update on the implementation of the Directive as regards European Reference Networks (ERN). A board of Member States was constituted on 5 December 2014 and rules of procedure adopted. The call for networks will be launched in December 2015: the assessment process will take place around March – May 2016 and in July 2016 decisions will be pronounced. Members stressed that **whatever grouping** is chosen, it must be carefully explained that this is just to provide guidance and direction and to avoid any overlap between potential ERNs: this is highly important as there will be a limited number of ERNs. This will help avoid overly narrow ERNs which may leave the most vulnerable, who are the most at need, without a structure regrouping expertise across Europe. It was highlighted that in the EUCERD Recommendations for Centres of Expertise, **the role of patients at the national level is clearly stated;** that they should get involved in the governance of the centre at national level. Patients will thus be involved at the national level.

COMMENT EAP May 2015 ***on grouping***
The system of the grouping comes with challenges for many rare disorders.
How would it work in practice when manifestations even within one family vary such as for example:
Neurofibromatosis *(1:3000)*
-Optic glioma (rare eye disease/ rare cancer)
-Pheochromocytoma (rare endocrine disorder / cancer)
- Skin neurofibroma (rare skin disorders)
- Learning problems (intellectual disabilities)
22qdeletion syndrome *(1:4000- 5000)*
- Cleft palate (rare head and neck disease)
- Cardiac defect (rare cardiac disease)
- Developmental and psychiatric disorders (brain and intellectual disabilities)
Stickler syndrome (*1: 7500- 9000)*
- Cleft palate (rare head and neck disease)
- High myopia (eye disorder – expertise of ophthalmologist)
- Hearing loss (hearing disorder – expertise of audiologist)
- Arthritis, scoliosis (orthopeadic disease)

B **- Interoperability and standardisation: connecting eHealth services**
The different degrees of interoperability: organisational interoperability; semantic interoperability, syntactical/technical interoperability, legal interoperability; were defined and the importance of effective governance stressed. The Health and Well-being unit of DG Connect can support within Horizon 2020 Research and Innovation initiatives for around 100M€ per year. A number of important projects promoting interoperability which are relevant to the field of health supported by the DG Connect were cited, including Antilope (advancing eHealth interoperability), EPSOS (interoperability of eHealth records), Expand (expanding health data interoperability services) and Sens (interoperable public services infrastructure).
Discussion: In Isreal there is an intergrated IT system involving primary care, including preventive medicine, and hospital care. ICD 9> 10 are implemented. Czech Republic electronic systems are used for statistics. The

government has asked input of all scientific societies for their needs of eHealth. This should be simple , with uniform codings. As Israel and the CzechR both use the same codes (ICD) these countries could compare data. Problem is the psychiatric diseases , because they are not open for all health care providers. The collected data are anonimized.
In Germany is the problem of data protection. There is a registry for cancer. Not all clinicians want to give away their data. There may be reluctance to get insight finance. And rare disease centres (specialists) are competing. Computers are not 100% secure.
Recommendation: Importance of integrated EHealth : epidemiological data, illustrate need of workforce; should benefit the patient. It should be defined what data are collected and why.

C. The Expert Group were encouraged to give their ideas on how to improve enrolment and to share experiences concerning initiatives that could help increase **participation in paediatric trials**.
The European Commission and the European Medicines Agency would be interested to learn the perspective of experts and organisations acting in the field of rare diseases, given that a considerable percentage of pediatric diseases are rare diseases. To this end, we would be grateful if you could consider the following questions in order to facilitate the discussion (you can also send us your input in writing in advance of the meeting):
The recruitment of children in clinical trials is low, what do you think / know is the reason for this?
- Recruitement of patients is extremely difficult due to regulations
- Reimbursement does not finance the structures needed
- The academic needs to deliver output on science , participation in trials is nor rewarded
- Pediatricians in academia need to compete with all other mainly 'adult' specialist
- The benefit for the child opposed to the group benefit should be considered
- For children harm (blood checks, etc) may exceed direct benefit, children cannot decide for themselves
- Pediatricians are expected to work as charities, and do all for free for the sake of the sick children
- Parents of RD patients do not aways agree to their child participation

Conclusion (after discussion at the General Assembly):
<u>We recommend independent funding for Pediatric Research and child health specially with regards to Medicine in children.</u>

D - Palliative Care in the European Union: Follow-up to informal Council discussions and proposals for a European Union framework of action: The Commission presented the informal discussions to date at the Council and proposals for a EU framework of action in the area of palliative care.
This issue has been discussed at the Cancer expert group and in other projects in other disease area.
A common approach to this question across different disease areas is favoured and the feasibility of an EU framework for palliative care considering the pediatric side of the issue and the perspective of healthy aging:
Yes , there sould be special guidance for palliative care for children.
Note: Archives of Diseases of Childhood just published an issue on this subject
Ref: Archieves of Diseases in Childhood. May 2015 Volume 100 Supplement 2
Making decisions to limit treatment in life – limiting and life- threatening conditions in children.
A framework for practices.
Conclusion (after discussion at the General Assembly):
<u>The EAP welcomes the initiative to bring this important issue on the European agenda.</u>
<u>We suggest special attention for training in pediatric palliative care for care-givers for children with potential lethal diseases as one of the major issues.</u>

E. In answer to the draft on social care the following respond was suggested:

- *Convention of the Rights of the Child* and the *Convention of the Rights of Persons with a disability* high light how children with disabilities have the same rights as other children. They are mutually reinforcing and together provide a framework for a growing synergy between key **human right** instruments
- For all children **early childhood** provides an important window of opportunity to prepare the fondation of life long learning and participation.
- Children with diabilities and their familie have ordinary needs and must have access to the **mainstream programmes** and services. Early childhood intervention should be integral part of existing health, educational and social systems.
- A disability or developmental delay should be addressed properly independant of the **diagnosis** of a (rare) disaese.

- The **identification of children with developmental delay or disabilities in primary care** needs support in the development of policies, strategic planning, and service planning.
- **Primary care pediatricians** are experiences in normal child development, identification of delayed of disabled children and family support. Training of pediatricians in secondary and tertiary care hospitals is base on pathologies split in the different specialist.

The medical home (or chronic care) concept is designed for a **collaborative management** approach at the primary health care level involving patients, their families, and other health care providers to reduce the burden of disease for the child, its family, and the community.

- **Coordinated and sustainable response** is required to preserve pediatric training and experience on normal growth and development as well as coordination of care of children with disabilities before primary care pediatricians disappear from societies.

Comment was discussed at the General Assembly and approved (see addendum to the minutes)

4. Projects: FP7- Rare Best Practices : www.rarebestpractices.eu
- Help to appraise guidelines with the AGREE method: there will be a workshop on the AGREE method during the EAP congress september 2015 in Oslo

5. Any other bussiness and proceedings: RBP session at EAP congress in Oslo September 2015

Present:

Abdulah Shamsah	Kuweit
Arunas Valiulis	Lithuania
Carsten Lincke	Netherlands
Gabriella Kubatova	Czeck Republic
Kaja Julge	Estonia
Kvetoslava Pruchova	Slovakia
Liesbeth Siderius	Netherlands
Manual Katz	Israel
Peter Hoyer	Germany
Sigita Burokiene	Lithuania
Tarek Khalefih	Canada

LS June 15th 2015