Minutes Rare Disease WG
(EAP, UEMS Section of Paediatrics)
Dublin, Friday June 3rd 2016,
WG Rare Diseases 9:00- 10:30 _Davenport Hotel Dublin

1. Attendance and apologies: no apologies

2. Minutes Winter Meeting Brussels, 2015 (january 2016- attached): no comments

3. Report from the EU Commission Expert Group on Rare Disease (april th5-6 2016) *Liesbeth Siderius*
New recommendation on social services:
http://ec.europa.eu/health/rare_diseases/docs/recommendations_socialservices_policies_en.pdf
The recommendations were reported.

4. Report from the Meeting of the Advisory Technical Group for the EU Platform on Rare Diseases Registration, Luxembourg, 7 April 2016 and First Interoperability Workshop organized in the framework of the EU Platform on Rare Diseases Registration, Luxembourg, 7-8 April 2016. *Liesbeth Siderius :*
Welcome European Commission *;* EU platform Rare Disease registration *;* Advisory technical group Interoperability needs – expected support and tools to be provided by the platform; EU projects Identification of concrete steps:
No clear steps have been proposed. As EAP representative the need of independent collections with international IT standards to support: shared community care; individualized, preventive medicine; medical research and quality improvement; enable epidemiological functions as well as routine health statistics; and determinate costs and better management of public and private payer funds.

5. 'Rare Diseases in Ireland, the challenge of delivery of care in a small member state'.
Prof. Eileen Treacy, MD, FRCPI, FRCPC, FCCMG
National Lead, Rare Diseases Clinical Programme, *Clinical Professor- Inborn Errors of Metabolism, Trinity College Dublin, Clinical Professor-University College Dublin, National Centre for Inherited Metabolic Disorders Children's University and Mater University Hospitals*

Ireland has an overall young population: 22% is under 14 years old. There is no national rare diseases registry. There are 16 smaller registries. Ireland participates in EUROCAT. There is a relatively higher incidence of birth defects documented in Eurocat (prenatal termination not legalized in Ireland). The national newborn screening programme has identified a higher incidence of metabolic diseases such as PKU and Galactosaemia. Cystic Fibrosis is not a rare disease in Ireland. Ireland has a distinct endogamous population: 'Irish travellers' who constitute approximately 1% of the total Irish population. In this population specific rare recessive conditions are relatively common such as galactosemia, Fanconi anaemia, MPS1 and anophtalmia.
The Irish national RD (2014-2018) plan contains 48 recommendations. One recommendation is the development of a National Clinical Programme for Rare Diseases and the opening of a National Rare Diseases Office, (opened in June 2015). One requirement/aim of the office is to provide trustworthy information, (as determined by a HSE 2012 Public Consultation and a study funded by the SAOIRSE Foundation).
The Rare Disease Office provides a website for patients, families and professionals to include General Practioners (www.rarediseases.ie).
The provision of care for Rare Diseases is provided through a number of Centres of Expertise and affiliated smaller centres.

Family carers Ireland
Catherine Cox : Mission 'to benefit the community by supporting and promoting the health, well being and quality of life of family carers and those for whom they care; to facilitate the affiliation of groups and group members representing family carers throughout Ireland and to liaise with, support and take counsel from such groups. Family Carers Ireland believes that: Care in the home makes a critical contribution to the welfare of society and this fact needs to be widely understood and acknowledged; Family carers deserve to be enabled and supported in their role and, in particular, to be accepted as an important partner of the state in the provision of clinical and social care; Supports and services for family carers should be accessible, sustainable and responsive to individual circumstances.

(1) FCI have over 130 carers groups across Ireland connected with us through membership and/or resource centres
(2) Many of these groups come together through commonalities such as the condition of the person they are caring for, their geographic location, the carers own age (young carers) or gender (male carers groups)
(3) As our organisation is for **all** family carers regardless of the condition of the cared for person it makes us unique and accessible to all carers including those caring for a loved one with a rare disease.
(4) These carers are a particularly vulnerable group due to lack of peer support, information and emotional support particularly at diagnosis stage.

and Johanne Powell Tells the story of her now adult daughter with a ring 8 chromosome abnormality. How she was informed on all the possible features and hardly understood what that would mean in real life. She advocates for involvement of families on all issue's concerning rare diseases. We are partners: the families have the experience the doctors the knowledge.
Discussion: more info in different languages; needed is the mix of patient expertise and professional science; examples on how this could work: the Duchenne pathway.
Still RD's are approached silos arranged by body systems and the specialism involved, a more holistic approach is needed. Other issue is DNA test availability in low economy countries.

6.Projects: FP7- Rare Best Practices : www.rarebestpractices.eu
Liesbeth Siderius on behalf of Harbour Jenny (HEALTHCARE IMPROVEMENT SCOTLAND);Ritchie Karen (HEALTHCARE IMPROVEMENT SCOTLAND); and Jorna Thomson (HEALTHCARE IMPROVEMENT SCOTLAND)
Power point presentation on results of search and AGREE method.
Results and report of the General assembly April 28-29th Tenerife: the project is in its last year. There is still an urgent need of more people to help with the appraisal of guidelines:

- Limited time remaining to expand the collection (Dec 2016)
- More volunteers needed to search for guidelines and appraise guidelines using AGREE II
- If you would like to contribute, contact:

Jenny Harbour Healthcare Improvement Scotland: jenny.harbour@nhs.net

7. Discussion : most of the discussion was under point 5: illustrating the need of input of carers and patient organisations

Guests:

Catherine Cox	Ireland
Johanna Powell	Ireland

Present:

Max Zach	Austria
Francis Crawley	Belgium
Peter Hoyer	Germany
Eileen Tracey	Ireland
Martin White	Ireland
Manuel Katz	Israel
Iveta Dzivite-Krisane	Letvia
Arunas Valiulis	Litouania
Liesbeth Siderius	Netherlands
Arthur Azur	Poland
Inez Azevedo	Portugal
Ana Neves	Portugal
David Neubauer	Slovenia
Jernej Zavrsnik	Slovenia

LS Concept June 25 th 2016