



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



Draft Minutes WG Rare Disease WG

Friday May 12th 2017 9.00- 10.30
Vilamoura, Algarve, Portugal

1. Attendance and apologies

2. Minutes Winter Meeting Brussels 2016, no remarks

3. Report UEMS Multidisciplinary Joint Committee Rare and Undiagnosed Diseases

The UEMS sets standards for high quality healthcare practice that are transmitted to the Authorities and Institutions of the EU and the National Medical Associations. An UEMS Multidisciplinary Joint Committee addresses a field of a multidisciplinary nature. The MJC RUD is established to bridge the gap between medical specialist societies and to facilitate high quality multidisciplinary care for the persons with a rare or undiagnosed condition.

MJC RUD has identified steps in the frame of the establishment of 24 European Reference Networks:

- harmonization of a common core training in principles of rare diseases (such as early recognition, access to diagnostic test, multidisciplinary care and societal expectations)
- provide CME accredited training programs at conferences and by E learning

4. Report on the 3rd European Reference Network Conference in Vilnius, March 9th 2017 24 thematic European reference networks where established.

Jean Claude Juncker, President of the European Commission mention in his video mentioned that many rare disease affect children. The aim of the ERN's is to assure better diagnosis and care for persons with a rare disease. Made possible by exchange of data between care providers, data travel not the patient, says Vytenis Andriukaitis, Commissioner on Health & Food Safety.

Christopher Fearne, Minister of Health Malta addressed the importance to include small patient groups in small population countries.

Video's en raports are at: https://ec.europa.eu/health/ern/events/ev_2010309_en

And the EAP twitters: twitter.com/euracademyaeds

5. "Rare Diseases Care Organization in Portugal"

by Maria do Céu Machado,

Vice President of the National Council for Health; Member of the National Bioethical and Life Sciences Council in Portugal; Vice President of the European Federation of Medical Academies and president of INFAMED

Portugal has been reducing its infant mortality rate significantly. From 55 in the seventies now below 3 about equal to Sweden. Portugal is now in the top 10 for newborns and children health.

1,8/1000 liveborns (9^o in 2012) Decrease 74% (1990 to 2012).

There is a national program for integrated health care for children. Transition into adult care is still underdiscussion.

The **Portugese national program for rare disease** (2008) aims to improve quality and safety of care

Specific objectives are to create a national registry; to improve the access to adequate care and to promote

integrated management; orphan drugs innovation; Training and research.

Rare Disease integrated management : Strategic priorities – To develop and improve (2015-2020):

- ❑ Healthcare Coordination
- ❑ Early diagnosis access
- ❑ Access to therapeutic innovation
- ❑ Clinical and epidemiological data
- ❑ Training and research
- ❑ Social integration and citizenship
- ❑ Patient and family involvement

Parliament recommendation to the government

To create a **RARE DISEASE (PERSONAL) CARD**

To supply all units with the equipment to print and read the card

To have a previous permission of the National Committee for Data Protection

To implement the strategy within one year.

The rare disease personal card is :To ensure the access to relevant information

Clinical data; Emergency recommendations (savelife); To improve integrated and continuous care;

To improve chronic disease management; Avoiding delay, error or inadequate intervention; To facilitate referral to reference centers.

Between February 2014 and april 2017 there have been 3785 cards requested.

SNIPi (National System for Early Intervention) - Lisbon Region 10 339 (3,7% population) children age 0-6 where elected. In the vision on long term investment, avoiding to focus on immediate cost.

Discussion:

In Ireland persons over 16 year can not enter pediatrics. The importance of collaboration with internal medicine is mentioned. In Portugal the CF centres are an example of intergration between child and adult health care. In the past most children died before they reached adulthood.

The personal card is part electronic part still on paper. Rare diseases can be identified by the orphacode. Not all patients want a personal card. 85 different diseases are registered.

Rare disease have to be build into a sustainable health care system. Polititions need to be aware and pharmacy can be a partner. How to make a step toward a european rare disease card (as in Italy). A focus document on a rare disease personal card could be helpfull to spread around in different european parties. June 18-19 will be a meeting of health ministers.

6. General discussion activities in the field of rare diseases

- results of questionnaire

A survey monkey questionnaire on rare diseases was send out to pediatricians that have been noted to have interest in this field in the past 6 years. The opinions of 23 respondents, in 16 different countries on 10 questions were reported.

The final question

What steps are necessary to extend and share knowledge in the field of rare diseases? In my daily practice the care for children with a rare disabling condition (more than one option possible) results:

- 1) 86 % a multidisciplinary approach would improve care
- 2) 69 % the availability of more information on rare diseases would enhance diagnosis
- 3) 56 % the availability of (international) guidelines would improve care
- 4) 52 % the availability of diagnostic tests would enhance diagnosis
- 5) 47 % membership of a European Reference Network would improve care

6) 8 % stated that at present the care is sufficient

- new activities:

On Sunday October 15th during the EAP Congress and Mastercourse in Ljubljana, Slovenia, there will be a round table conference on IT, primary care and rare diseases.

Participation will be free of charge.

Attendance:

Maria Machado	Portugal
Christine Aebi	Switzerland
Peter Altorjai	Hungary
Ines Azevedo	Portugal
Sita Burokiene	Lithuania
Peter Hoyer	Germany
Jose Manual Lopez dos Santos	Portugal
David Neubauer	Slovenia
Ana Neves	Portugal
Vladimir Pilosoff	Bulgaria
Paul Soler	Malta
Martin White	Ireland
Liesbeth Siderius	Netherlands

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