



15th of May

Minute of Rare Disease WG (EAP, UEMS Section of Paediatrics)

Rare Disease Working Group

Friday May 11th 14.00-15.30

Prague

1. Attendance and apologies

Dodge John, Cooley Carl, Stefanidis Constantinos, Einarsson Ingelfur, Wierzba Jola, Ramos Feliciano.

2. Minutes Meeting December 2011

No comment

3. Report on First International Workshop Clinical Practice

GUIDELINES on Rare Diseases; ROME, FEBRUARY 2012

EAP was invited to be in the meeting organized by Italian Public Health Organization. Liesbeth Siderius were representing EAP in the workshop.

The workshop was attended by health technology assesment experts, public health representatives, and patient organisations, among others.

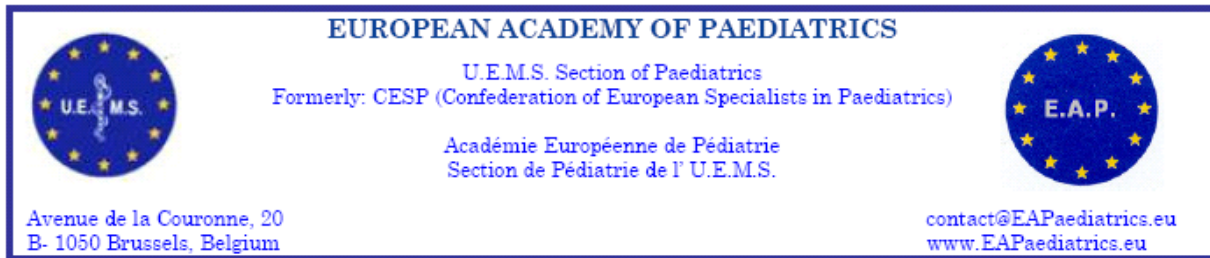
Clinical Practice Guidelines are needed to harmonize the clinical practices all over Europe.

Guidelines on rare disease are sparce. At present there is a tendency that larger countries develop their own national guidelines. Small countries will not be able to do the same.

How can we collect the existing guidelines or how can we create them and / or harmonize them among Europe?

We need a guideline to create guidelines for rare disease: the amount of guidelines to be written is enormous, **but there are common features to address.** Guidelines main objectives should be:

- Contribute to reduce fragmentation of care and
- help coordinate the care of rare diseases
- help the individuals getting the reinboursment of their rare disease.



The countries have the same definitions of rare disease. The European Committee recognized terminology (1:2.000).

Lack of evidence based is a problem to get guidelines that could be accepted by insurance companies. Most of paediatrics guidelines are moving from evidence based to consensus based.

A suggestion from Norway Mrs Elisabeth Siebke: we have booklet for certified rare diseases, and small group disease. We can take them and translate them.

Other suggestion: start with one rare disease quite known all over Europe like cystic fibrosis.

Hoyer Peter from Germany propose to start with a list of common problem that has the rare disease. And some instruction to deal with this problems.

What are the characteristic of all rare disease:

1. Late diagnosis
2. Parent need of reliable information
3. Fragmented care
4. Problem of reimbursement of multidisciplinary care
5. Coordination of care: in institutions as well as ambulatory care.

We can start from this point to create a matrix for guidelines addressing the above common problems.

4. Training in Rare diseases, options:

Rare disease often not recognized. **Training is crucial.**

Who should be trained?

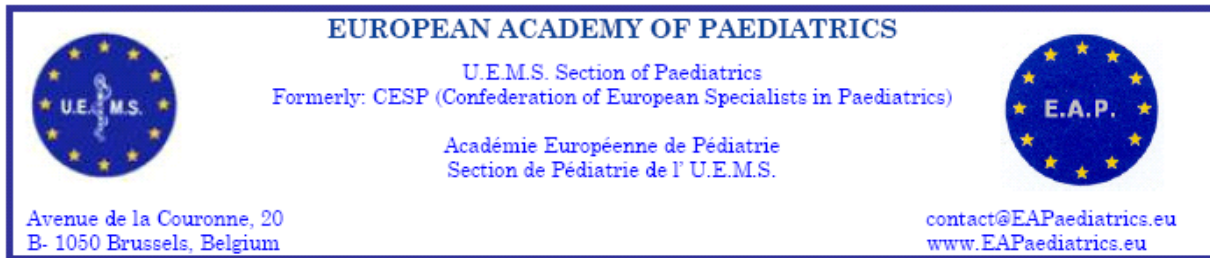
- General Paediatrician?
- And/ or Paediatric Specialist?
- Or E-learning for both?

You can find most information through 'Google', but it is difficult to find the good information.

We suggest to create a UEMS accredited e-learning course to be provided by EAP for all interested.

5. Projects presented for EU funds

- With a project on »Harmonisation of standards of care applied« we applied for EU funds. The project was rejected, while an Italian group succeeded in something very similar. EAP is now collaborating on this project with the italians.



- Another EU proposal was submitted for EU funds in the program Health. The proposal is based on the idea to create in Europe a web-portal similar to the US portal www.medicalhomeportal.org/

6. Fothcoming Meetings

- Global Child Health meeting (Consensus in Paediatrics) May 17-20 2012
- EURORDIS meeting May 23-25 2012

7. Discussion

Rare disease definition

Prof. Tenore made some considerations on the definition of rare diseases.

New born screening are effective if:

- The disease is quite frequent
- You can change the evolution of the disease if you recognize it in time
- The cost of the screening has to be not much compared to the cost of the disease management

Because new born screening are made principally to save community money and they should be effective financially.

But how we define »frequent«

More than 1 out of 15.000 was the previous definition.

Now it is 1 out of 2.000 and this make most of the diseases in childhood rare.

A good starting definition could be to consider rare diseases with incidence 1 or less out of 10.000.



The change in the definition of rare diseases has been made for political reason. Now there is governmental attention on these diseases. So if it is defined as rare it is more likely to access to fouding.

National Plan

Now in Europe every country have to prepare a national plan for rare disease management.

Discussion arouse on how each country present at the meeting is preparing the national plan.

- Norway and Sweden are doing it with the involvement of the Paediatric Associations
- Germany, strange situation
- Poland, only paediatric genetist are involved in it
- Lithuania now all in the hand of the Ministry of Health. Too much burocracy. But election are near.

	EUROPEAN ACADEMY OF PAEDIATRICS U.E.M.S. Section of Paediatrics Formerly: CESP (Confederation of European Specialists in Paediatrics) Académie Européenne de Pédiatrie Section de Pédiatrie de l' U.E.M.S.	
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Thank you everybody for being present to the meeting.

People in the meeting:

Chair: Liesbeth Siderius – The Netherlands

Participants:

- | | |
|--------------------------|----------------|
| 1. Azavedo Ines – | Portugal |
| 2. Frecerova Klara – | Slovakia |
| 3. Hoyer Peter – | Germany |
| 4. Hrdlicka Renè – | Czech Republic |
| 5. Marquis Ana- | Portugal |
| 6. Mazur Arthur – | Poland |
| 7. Neubauer David – | Slovenia |
| 8. Palm Palm – | EPNS |
| 9. Siebke Elisabeth – | Norway |
| 10. Szczepanski Tomasz – | Poland |
| 11. Tenore Alfred – | Italy |
| 12. Valiulis Arunas- | Lithuania |
| 13. Wettergren Bjorn - | Sweden |