



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



Draft of January 1th 2014

## Minutes of Rare Disease WG (EAP, UEMS Section of Paediatrics)

**Rare Disease Working Group, Saturday 14 december 2013**  
Brussels

### 1) Attendance and appologies

John Dodge, (UK), Cooley Carl (US), Stefanidis Constantinos (Greece), Einarsson Ingelfur (Iceland), Jola Wierzba (Poland), Ramos Feliciano (Spain) , Zoran Gusev (Macedonia), Larissa Prikhodina (Russia)

### 2) Minutes Meeting December 2012:

Not available.

### 3) Guidelines

A. Arnold Oranje, dermatologist, Rotterdam, The Netherlands:

- Mastocytosis: As an example of a rare disease with a distnished benign course in children. The type of skin manifestations indicate a good or less better prognosis. In the adult form bonemarrow investigations are recommended, while in children almost never. Therefore the WHO classification is not applicable for children. A International management guideline is available.

B. Projects: FP7- Rare Best Practices : [www.rarebestpractices.eu/home](http://www.rarebestpractices.eu/home)

Update Meetings 2013:

- Rome (February) 'Kick of' meeting
- London (May) - journal discussed : first issue December 2013 : [www.rarejournal.org](http://www.rarejournal.org)
  - guideline database
  - pilot guideline (T. Sejersen, Sweden)
- Freiburg (October): methods according the GRADE

Which diseases to select? Suggestions? : List of proposed diseases discussed; international guidelines are preferred; EAP can act as a platform (at the website) and collect guidelines especially from member societies.

The Rare Disease WG Contributes to meetings in 2014

20-21 March Rome, Italy: General Assembly Rare Best Practice

28-31 May, Rzeszow, Poland: Spring meeting EAP: update

17-21 October EAPS Barcelona, Spain : workshop/ session: "Rare diseases, common in paediatrics"

### 4. ICF-CY codes (WHO) for rare diseases

Huib ten Napel , WHO-FIC Collaborating Centre, Bilthoven , The Netherlands

The ICF- CY is member of the WHO family of international classifications (WHO-FIC). It offers a more detailed description for childrens functioning properties in an international standardised terminology and framework. This can support the collection and exchange of data concerning Rare Diseases in children, required for research and policy making.

### 5. Comment on UNICEF/WHO discussion paper on Early Childhood Development and Disability(draft)



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The paper has been circulating since december 2012. In the ethics group Francis Crawley made some editorial corrections. The adherence of the EAP will be asked at the general assembly.

Coordinator: Liesbeth Siderius (Netherlands)

Present:

Radovan Bogdanovic (Serbia)

Marieke Emonts (Netherlands)

Hand Jurgen Dornbusch (Austria)

Ilze Grope (Russia)

Peter Hoyer (Germany)

Karoly Illy (Netherlands)

Gabrieza Kubatovag (Czech Republic)

Carsten Lincke (Netherlands)

David Neubauer (Slovenia)

Arnold Oranje (Netherlands)

Vladimir Pilosoff (Bulgary?)

Ninel Revenev (Moldavia)

Tom Siris (Norway)

Liis Toome

Arunas Valiulis (Lithuania)

Leslie Villalever (Netherlands)

Max Zach (Austria)

Dd January 1th 2014