

# Rare Diseases Working Group



Tartu, Estonia, September 2014

# Rare diseases WG

- Established in 2011
- 2012/13: Survey on the awareness of RD in Europe
- 2014: Position Paper of the Rare Diseases working group of the European Federation of Internal Medicine

# Rare diseases WG

- 2014: EFIM appointed as one of the 4 representative of the professional organisations and scientific societies in **Commission Expert Group on Rare Diseases of the EU Commission**



DIRECTORATE GENERAL  
HEALTH AND CONSUMERS

# Commission Expert Group on Rare Diseases of the EU Commission



- Instituted by the Commission Decision 2013/C 219/04 of 30 July 2013
- Replace the EUCERD



# Commission Expert Group on Rare Diseases of the EU Commission



## Mission of the group:

- assist the Commission in the drawing up of legal instruments and policy documents, including guidelines and recommendations;
- advise the Commission in the implementation of Union actions and suggest improvements to the measures taken;
- advise the Commission in the monitoring, evaluation and dissemination of the results of measures taken at Union and national level;
- advise the Commission on international cooperation;
- provide an overview on Union and national policies;
- foster exchanges of relevant experience, policies and practices between the Member States and the various parties involved.

# Commission Expert Group on Rare Diseases of the EU Commission



## Composition of the group

- Member States' competent authorities (28)  
+ Icelandic Ministry of Welfare, State Diagnostic and Counselling Centre; Norwegian Ministry of Health and Care Services
- Patients' organisations in the field of rare diseases (4)  
EURORDIS – European Organisation for Rare Disease; The Network of European Federations of rare diseases; EGAN – European Genetic Alliances' Network; The Network of National Alliances of rare disease patients' organisations
- European associations of producers of products or service providers relevant for patients affected by rare diseases (4)  
European Association for Bioindustries – EuropaBio; European Biopharmaceutical Enterprises; The European Diagnostic Manufacturers Association; The European Social Insurance Platform
- European professional associations or scientific societies acting in the field of rare diseases (4)  
**The European Federation of Internal Medicine**; European Society of Human Genetics; European Hematology Association; European Academy of Paediatrics
- individual experts (4)  
Domenica Taruscio (I); Katharine Bushby (UK); Milan Macek (CZ); Ségolène Aymé (FR)

# Commission Expert Group on Rare Diseases of the EU Commission



## Activities

- EFIM is the contact point for Professional Societies
  - The European Federation of Internal Medicine
  - European Society of Human Genetics
  - European Hematology Association
  - European Academy of Paediatrics

# Commission Expert Group on Rare Diseases of the EU Commission



## Activities

- European Reference networks status of the implementation of the Art 12 of the Directive 2011/24/EU on patients' rights in cross-border healthcare
- Rare diseases codification
- New Joint Action on rare diseases
- Rare diseases registries
- Research infrastructures in the field of RD research

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# Proposed activities for the EFIM Rare Disease WG

## Proposed activities

- EU survey on how internists deal with rare diseases
- EU School for rare diseases
- Childhood to Adulthood transition project (with EAP)



**EFIM**

European Federation of Internal Medicine



# Proposed activities for the EFIM Rare Disease WG

## Proposed activities

MARIE SKŁODOWSKA-CURIE  
ACTIONS



EFIM

European Federation of Internal Medicine



# Rare Diseases Working Group



Thank you