

## **Report of the 1st WG on Registries and Natural History Meeting**

Dublin, Burlington Hotel

15<sup>th</sup> April 2013

### **Participants**

Dr Anna Ambrosini, Milan, Italy  
Mr Kyle Brown, San Mateo, USA  
Dr Josh Burns, Sydney, Australia  
Ms Liz Donohue, Sioux Falls, USA  
Dr Mike Innes, Calgary, Canada  
Ms Virgine Picard, Paris, France  
Dr Manuel Posada, Madrid, Spain  
Prof Bernard Zabel, Freiburg, Germany  
Dr Cornelia Zeidler, Hannover, Germany  
Dr Rumén Stefanov, Plovdiv, Bulgaria, ISC Member  
Dr Domenica Taruscio, Rome, Italy, ISC Member  
Ms Samantha Parker, Puteaux la Défense, France, ISC Member

### **Introduction**

- ▶ Introduction from WG members
- ▶ Explanation of IRDiRC governance and structure
- ▶ Objectives of the WG and reporting rules

### **Role, Mandate and Focus of the WG**

Following IRDiRC policy:

#### **a) The mandate of IRDiRC Working Groups is to:**

- ▶ Cooperate to ensure synergies of all research projects within the scientific area of the working group, by exchanging results, expertise, experiences and information
- ▶ Identify opportunities and gaps in RD research (contributions to the IRDiRC road map)
- ▶ Point out the problems and difficulties of RD research that prevent or slow down progress towards diagnostics and therapies
- ▶ Suggest new topics for RD research and advise on the format of future projects
- ▶ Analyze and propose standards (e.g. quality standards, performance indicators) for RD research
- ▶ Report on the implementation IRDiRC policies and guidelines in RD research projects and propose changes where necessary

- ▶ Report on their findings to one or more Scientific Committees

**b) Registry and natural history Working Group shall cover the following topics:**

- ▶ CDEs: need for interoperability / MDS minimal data set to be defined
- ▶ Sustainability plan enabling patients for self entry and PPP interoperability and clustering of registries
- ▶ Quality assurance and data integrity (linked to the core MDS)
- ▶ Standardization of outcomes measures
- ▶ Linking to medical records

**Goals for the 1st WG meeting:**

- ▶ Discuss and nominate Chair and co-Chair
- ▶ Discuss and comment on topics and goals of the WG
- ▶ Identify needs, gaps and opportunities

**Discussion**

Elections:

- ▶ Domenica Taruscio has been elected chair of the working group.
- ▶ Liz Donohue volunteered to be co-chair of working group.

Discussion included CDEs / MDS and the need for interoperability, Global Rare Diseases Repository project (GARD) and other funded projects (e.g., RD-Connect, Epirare, GRDR).

Quality assurance and data integrity (linked to core data set) were subsequently discussed. Additional ideas that were also discussed extensively:

- ▶ Sustainability plan enabling self reported data and PPP/interoperability and clustering of registries. Funding will be a major issue and may affect how much data to collect.
- ▶ Global unique patient identifier ID. Standardization of outcomes measures.
- ▶ Medical Records.

Participants proposed potential collaboration with other Working Groups. Other WG members could be joining the next meeting of this group. A possible way to approach this task could be to initially establish principles, and next to collect and share existing tools and/or guidelines, and focus on implementation.

A follow-up phone call was planned to take place before the summer.