

## Meeting report series

# Report of the 1<sup>st</sup> Teleconference of the WG on Biomarkers for Disease Progression and Therapy Response

5<sup>th</sup> December 2013

## Organization

Organized by: Scientific Secretariat

Hosted by: Teleconference

## Participants

Dr Gillian Butler-Browne, Paris, France

Dr Giles Campion, Leiden, Netherlands (member of the TSC)

Prof Alessandra Ferlini, Ferrara, Italy

Dr Marlene Haffner, Rockville, USA

Dr Barbara Cagniard, Scientific Secretariat

Dr Virginie Hivert, Scientific Secretariat

Dr Sophie Höhn, Scientific Secretariat

## Apologies

Dr Spiros Vamvakas, London, UK

Dr Marc Walton, Silver Spring, USA

Dr David Wishart, Edmonton, Canada

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## Agenda

- 1) Welcome and Introductions
- 2) Role and support of IRDiRC secretariat
- 3) Review of WG Briefing Document
- 4) Review of the list of projects related to Biomarkers funded by IRDiRC members
- 5) Election of the WG chair
- 6) WG Deliverables and timing for next call

## REPORT

### 1) Welcome and Introductions

Each participants of the WG teleconference introduced themselves. The participants have all a specific interest in biomarkers and related appropriate endpoints, either in the academic or industrial sectors, with a majority of them working in the field of rare neuromuscular diseases.

### 2) Role and support of IRDiRC secretariat

The project manager of the IRDiRC secretariat introduced briefly the team that is located in Paris. The team is composed by 4 people, including a project manager, a communication manager, an information scientist and an assistant. Support in kind is also given by people from the French Rare Diseases Foundation and from Orphanet. The role of the IRDiRC secretariat is to help organizing the meeting and teleconference of the Committees and Working Groups (WGs), to take the minutes and propose a report, but also to prepare documents upon request, including extraction of data from the Orphanet database or literature survey when needed.

### 3) Review of WG Briefing Document and 4) Review of the list of projects related to Biomarkers funded by IRDiRC members

#### General mandate and main tasks

The participants of the WG on Biomarkers have reviewed the general mandate and main tasks. The key points that have been identified are:

- ▶ Item 3: Point out the problems and difficulties in the scope of the WG that ultimately prevent or delay in developing new diagnostics and therapies
- ▶ Item 5: Cooperate to ensure synergies of all research projects within the scientific area of the WG, by exchanging results, expertise, experiences and information
- ▶ Item 6: Analyse and propose standards to be promoted (e.g. quality standards, performance indicators) for RD research

The main goal is thus to establish points to address and tasks to do after review of the elements for the gap analysis. So according to the participants, the first task is to go through the list of projects related to biomarkers to see if collaborations could be foreseen and suggested by the WG. To do so, biomarkers could be divided into several groups, by type of diseases for example. This would require involving people working in other rare diseases areas.

The list provided by the IRDiRC secretariat was just to present the projects funded by the IRDiRC members during the time period covered by the Consortium (from 2010). This list has to be enlarged to better represent the landscape in the field.

Additionally, one of the most challenging issues would be to discuss how to convert biomarkers, identified or used mostly in an academic settings, into surrogate clinical endpoints. Another issue has been raised, whether the WG should focus on biochemical biomarkers only. According to the group, the most important is the function, i.e., to show the link between a biomarker and a function, even if the link/pathway is still unknown. The participants agreed to concentrate on biochemical biomarkers but not imaging (except if it concerns a parameter that is measurable).

### **Specific topics to be addressed by the WG**

The members of the WG have then reviewed the list of specific topics, mainly items 2 to 5.

It has been mentioned that part of the discussion to be held is very close to the topics of the Diagnosis Scientific Committee working groups because biomarkers may become a tool for diagnosis. So a common conference call with WG from the DSC has to be scheduled.

It was also brought out that the gap analysis may include a questionnaire to be sent to the project leader of projects on biomarkers and related issues (as gene discovery, mechanisms definition etc) in rare diseases to ask them for key points to be further discussed or prioritized.

The item 4 “Interaction with regulatory agencies for qualification and validation of biomarkers to further accelerate orphan development” has been identified as a big issue especially in the US. Indeed, a biomarker may be specific of a drug and may also be measured by different ways in different labs.

### **5) Election of the WG chair**

Alessandra Ferlini has been elected as the chair of the WG on Biomarkers.

### **6) WG Deliverables and timing for next call**

#### **Summary of the discussion**

The mandate of the WG on Biomarkers has a broad scope which needs to work with a list of projects in the field as comprehensive as possible and representing the full EU/US picture, to be divided by diseases or groups of diseases. The most promising projects have to be identified in order to focus on them. The Duchenne Muscular Dystrophy history could be used as a paradigmatic flowchart of the necessary steps towards clinical trials.

Efforts have to be put on standardization of biomarkers for clinical trials:

- ▶ mainly low-, non-invasive markers, such as on biomarkers from blood and urine but not from tissue,
- ▶ also functional/readable biomarkers.

Natural history is also of high importance and a list of projects labelled natural history will also be proposed by the IRDiRC secretariat, in addition to the above mentioned extended list of projects related to biomarker and related research.

### **Tasks for the next meeting**

- ▶ Members of the WG will work on criteria and process to fulfil the regulatory requirements and on identifying scientific advice for biomarkers. Contact with EMA/FDA is foreseen.
- ▶ As mentioned above, the IRDiRC secretariat will provide a broader list of projects related to biomarkers extracted from the Orphanet database, and will also work on the Cordis and Clinicaltrials.gov website with the keywords 'biomarkers', 'natural history' and 'rare diseases' as to provide the WG members with a more comprehensive picture of the field. The list of flags used within the Orphanet database to label the research projects will also be provided.
- ▶ A topic has to be deeper explored, regarding how to find information on biomarkers when the trial failed.
- ▶ Harmonization between EU and US has to be kept in mind.