

Meeting report series

Report of the 3rd Companies Constituent Committee Meeting

Teleconference

March 6, 2018

Participants

Dr Mathew Pletcher, Roche, Switzerland (Interim Chair)

Dr Katherine Beaverson, Pfizer, USA

Dr Tim Considine, Recursion Pharmaceuticals, USA

Dr Madhu Natarajan, Shire, USA

Dr Tom Pulles, Ultragenyx, Switzerland

Ms Christine Cutillo, NCATS, USA

Dr Marlene Jagut, Scientific Secretariat (Sci Sec), France

Dr Anneliene Jonker, Sci Sec, France

Dr Lilian Lau, Sci Sec, France

Apologies

Ms Karian Aiach, Lysogene, France

Dr Andrea Chiesi, Chiesi Pharmaceutici, Italy

Dr Carlo Incerti, Genzyme, USA

Dr Ning Li,

Dr Robert Mashal, NKT Therapeutics, USA

Dr Brett Monia, Ionis Pharmaceuticals, USA

Dr Ellen Welch, PTC Therapeutics, USA

Dr James Wu, WuXi AppTec Co., Ltd., China

Agenda

1. Welcome new members
2. Review approved CCC activities – Document 1
 - a. Activity D: Natural history and registry (NH/R) platform for use in real world evidence (RWE) data collection

- b. Activity H: Background internal work on common knowledge base to drive rare diseases research
3. Discuss next steps
4. Official election of CCC Chair and Vice Chair
5. Any other business

REPORT

1. Welcome new members

The Interim Chair of the Companies Constituent Committee (CCC) welcomed the meeting participants, who each introduced themselves. Representatives present on the call were asked to introduce themselves.

Mathew Pletcher

- ▶ Acting Chair of CCC
- ▶ Head of Rare Disease Discovery at Roche, Switzerland

Katherine Beaverson

- ▶ Senior Director and Patient Advocacy Lead, Rare Disease Research Unit at Pfizer, US

Christine Cutillo

- ▶ Special Assistant to Dr Austin, National Center for Advancing Translational Sciences (NCATS), NIH, US

Tim Considine

- ▶ Senior Vice President, Strategic Development at Recursion Pharmaceuticals, US

Tom Pulles

- ▶ Head of Medical Affairs & Patient Advocacy, Europe at Ultragenyx Pharmaceutical Inc, Switzerland

Madhu Natarajan

- ▶ Research Therapeutic Area Head, Ophthalmics & Complement Biology at Shire,

2. Review approved CCC activities

Two CCC activities have been approved and included in the IRDiRC 2018 Roadmap, that resulted out of the Tokyo CCC discussion. These activities have been selected to together help drug discovery move forward.

2.1 Activity D 2018 - Natural history and registry (NH/R) platform for use in real world evidence (RWE) data collection

Activity D of the IRDiRC 2018 roadmap, “natural history and registry (NH/R) platform for use in real world evidence (RWE) data collection”, is an activity led by the CCC, but it will be jointly set up with assistance from the TSC and FCC, who both also suggested this activity.

Goals of this activity:

- ▶ Establish best practices: identify patient groups to pilot project
 - Better empowerment of patient advocacy groups to help provide data
 - Patient groups want to help provide NH data, but there is a lack of best practices in engaging patients for (long term) data collection
- ▶ Define standards: for collection of high-quality and interoperable data
 - Overcome the lack of standard-based data collection
 - Overcome the lack of standard to guide data collection
 - Overcome the lack of platforms to adequately record data
- ▶ Building an infrastructure backbone
 - Allowing for real-world data to fill in the backbone
 - Possibility to marry modeling of NH data with data collection in the ERN/ and US CRNRD
 - Possibility to marry academia, industry and patient advocacy groups
- ▶ Focus on building knowledge of diseases currently without available therapies
 - Allowing to get better natural history data
 - Opening up the space of R&D, due to a larger availability of data
 - Overcoming heterogeneity of knowledge
 - Being able to trace long-term impact, tracking patients over time
- ▶ Ideally based on existing case studies, covering different technologies and therapeutic areas
 - Learn from best-case examples, such as examples from DMD

2.2 Activity H 2018 - Background internal work on common knowledge base to drive rare diseases research

Activity H of the IRDiRC 2018 roadmap, “background internal work on common knowledge base to drive rare diseases research”, is the second activity led by the CCC. This will be a preparatory phase for the more extensive work that should be taken on by a postdoc in a later stage. It will allow the CC to decide what exactly they want to accomplish in the long term.

Goals of this activity:

- ▶ To better organize data and to make data available
- ▶ Possibility to generate knowledge away from the 10-15 core diseases everyone is working on and address the 6,950 diseases without activities
 - Establish a list of all rare diseases, including subtypes of a disease
- ▶ To improve prevalence numbers, allowing for better statistical data for each disease
 - Current numbers are often incorrect, underestimated, or pulled together from large generic data sets
 - Provide support to frequently cited statistics, e.g. number of diseases
 - Provide data based on recent diagnostics categorization
 - To allow for a better organization and integration of several large data sets, such as the 100K Genome Project, or deep sequencing efforts in Dubai

- ▶ System will enable new drug discovery
 - Highlight the next level of diseases with high potential for development
 - Academia can de-risk some studies, improving attractiveness for development

3 Next steps

3.1 Activity D 2018 - Natural history and registry (NH/R) platform for use in real world evidence (RWE) data collection

Discuss with Chair of TSC

- ▶ Set up call with Chair of TSC, on what he thinks are the next steps

Preparation of a 2-day workshop

- ▶ Build and discuss simulated cases
- ▶ Output: recommendations on how to build an ideal system for use in the real world
- ▶ Output: recommendations of standards and best practices in data collection
- ▶ Eventual, for long-term use: craft the infrastructure
 - This will require substantial resource to build and maintain
- ▶ Multi-stakeholder involvement

4 Official election of CCC Chair and Vice Chair

- ▶ The election of the CCC Chair and Vice Chair would be taking place based on the following procedure:
 - Host a call for nominations, in which we ask CCC members that are interested in the position of Chair or Vice-Chair to send their CV and a short paragraph stating their motivation (total 1A4 text) (1-2 weeks)
 - Sci Sec will compile incoming nominations (1 week)
 - Compiled nominations will be send it to all CCC members for an anonymous vote (1-2 weeks)
 - The Sci Sec will announce the selected Chair and Vice Chair shortly after the closing of the vote

5. Any other business

There is a new initiative, the Global Commission (<http://www.globalrarediseasecommission.com/>), set out to end the diagnostic odyssey for children with a rare disease. This Global Commission is a joint initiative of Shire, Microsoft and EURORDIS-Rare Diseases Europe.

- ▶ Follows a previous initiative of Shire and Global Genes
- ▶ Cross sector approach for rare disease diagnostics

- ▶ Still in its first year, so currently setting out goals and roadmap
 - Possibility to influence this initiative and direct their goals and work plan