Meeting report series

Report of the 1st ISC Working Group on Registries and Natural History teleconference

July 8, 2013

Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

Dr Domenica Taruscio, Rome, Italy, chair
Ms Liz Donohue, Sanford Research, USA, co-chair
Prof Odile Boespflug-Tanguy, Paris, France
Ms Anna Kole, Paris, France
Ms Samantha Parker, Puteaux la Défense, France
Ms Virginie Picard, Paris, France
Dr Manuel Posada, Madrid, Spain
Dr Rumen Stefanov, Plovdiv, Bulgary
Dr Cornelia Zeidler, Hannover, Germany
Dr Yaffa Rubinstein, Bethesda, USA

Dr Ségolène Aymé, Scientific Secretariat
Ms Roseline Favresse, Scientific Secretariat
Dr Sophie Höhn, Scientific Secretariat

Apologies

Dr Jan Friedman, Columbia, Canada
Dr Micheil Innes, Calgary, Canada
Pr Eugenio Mercuri, Rome, Italy
Pr Kathryn North, Sydney, Australia
Pr Bernard Zabel, Freiburg, Germany

REPORT

Introduction of the Scientific Secretariat

Following the welcome of the participants by the chair of the WG, the Scientific Secretariat was briefly introduced. The Scientific Secretariat is located in Paris and composed of four full time employees (project manager, communication manager, research scientist and assistant). Employees from Orphanet and the French Foundation for Rare Diseases also provide support in kind. The role of the Scientific Secretariat is to bring organizational support to IRDiRC Executive Committee, Scientific Committees and Working Groups by, among others, helping organizing meetings and teleconferences, writing the report of these meetings/teleconferences, and prepare any necessary documents upon request.

Issues and concerns for discussion

Cooperate to produce a well-curated list of all rare diseases:

- ▶ Should we consider the European list published as an Orphanet Report Series (monthly update) for adoption?
    - o It is impossible to have a unique list of rare diseases because of the different rare disease definitions existing in the world (different prevalence)
    - o List the different definitions for different countries or create a common definition for IRDiRC project accepted by the Executive Committee
    - o Explain the difficulties we are facing
- ▶ Agree on a definition to have the same list of rare diseases across IRDiRC members
    - o Definition for a single rare disease or a unique definition for rare diseases?
    - o Different legislations and definitions for rare diseases in different countries
    - o Do we have a need for a unique and well-curated list of all rare diseases?
- ▶ Encourage researchers to contribute to the update of the list
    - o Should we encourage researchers to contribute to that list?
    - o List of diseases, some of them rare, already exist such as the 11th version of the International Classification of Diseases (ICD 11). Orphanet has a lot of potential rare diseases already accepted by experts. Should we contribute to ICD11?

The goal of this working group would be to create a common definition for rare diseases which could be used by all countries and could standardized efforts. Having a unique definition for rare diseases and a unique list of rare diseases would be a big step forward, but it seems really difficult to achieve. Core of federal diseases in common to many countries could be a solution.

Promote a single set of standards for collecting, storing, annotating and communicating data:

- ▶ Review existing standards to precisely identify the areas for setting standards
    - o Common Data Elements by the Office of Rare Diseases Research (ORDR)

- o RD-Connect platform
- o EPIRARE platform
- ▶ Fund projects to reach consensus
- ▶ Disseminate the standards and make them mandatory to get funding

The creation of a set of Common Data Elements (CDEs) by the Global Rare Diseases Patient Registry and Data Repository (GRDR) and the Office of Rare Diseases Research (ORDR) was mentioned as a good start and a working document for IRDiRC set of standards. A validation of that document could be needed first. Adjustments will be required as many countries belong to IRDiRC. Many platforms such as RD-Connect and EPIRARE are working on the definition of the common data elements and further steps to collect data in a standardized way. This seems to be a good strategy to share the common data elements in different actions (national or European), and to try to describe a final list of common data elements as first steps. It could facilitate data collection in a standardized and meaningful manner, and facilitate harmonization between the many organizations collection patients information.

Contribute to the development and evolution of a set of standards for RD natural history studies:

- ▶ Review existing standards to precisely identify the areas for setting standards
  - o The National Database for Autism Research is a good example of standardization
- ▶ Fund projects to reach consensus
- ▶ Disseminate the standards and make them mandatory to get funding

The most difficult thing is that the IRDiRC set of standards will have to give rare disease natural history. It will require to:
- ▶ Define a set of common data elements for all rare diseases, and add this model for general rare diseases registries
- ▶ Identify specific experts for rare diseases that could standardized specific common elements for natural history of the rare disease, and share in a platform all the common data elements

Pursue the interoperability and harmonization between RD patient registries:

- ▶ Review standards used by existing patient registries
- ▶ Fund projects to reach consensus
- ▶ Disseminate the standards and make them mandatory to get funding

Interoperability is a definitional problem, not a technical challenge. If everybody used a common methodology to collect data, interoperability would be simpler. The biggest challenge is what people are collecting, how it is defined, and what consensus they have in order to do the interoperability. In GRDR program, they find the lowest common denominator and ask people to map their collected registry data to the common data elements. A simple questionnaire is sent to patients and then made public. Interoperability is simple enough that less sophisticated registries can implement technologies. The first step is to agree on the kind of data collected, the nomenclature and the common ontology.

The validation of the database is important. As many definitions exist for the same rare disease, a very simple definition of the symptoms should be included in the database.

Other issues

Global Unique Identifier (GUID)

The GUID aims to have a unique code for each patient around the world. The GUID can identify that it is the same person even if the information comes from different sources and registries. The information indicated in official documents that do not change (first name, last name, sex, country of birth) could provide minimal data set elements and allow creating a global unique identifier and connecting data with a specific person. Elements of the GUID are collected by a questionnaire on common data elements (ORDR). Several actions, mainly at European and American levels (NIH, Global Rare Disease Registry), could provide a GUID for registries and a platform for database research. There are about ten European projects that try to standardize data and interoperate (for example, Health European Care). But none of the European projects have the capacity to provide a unique GUID. Two experiences in USA are trying to follow the patient considering he is in one system. The NIH may invest in the creation of the GUID. International character set should be tested.

Main deliverables

- ▶ Collect short bio of all WG members
- ▶ Contact members unable to attend the teleconference to inquire about other issues they want the WG to discuss in September
- ▶ Diffuse documents that could help us to understand the CDEs from ORDR
- ▶ Circulate information about the National Database for Autism Research
- ▶ Provide additional information and documents on the GUID
- ▶ Collect and circulate information on the activity of the Ontology WG
- ▶ Add an additional point to IRDiRC Action plan, "n°20 - Develop metrics of progress and success"
- ▶ Send a doodle to plan the next teleconference to be held the last week of August or first week of September