IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 2nd DSC WG on Ontologies and Disease Prioritization teleconference

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Organization

Organized by: Scientific Secretariat
Teleconference

Participants

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Apologies

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REPORT

Update on Global Alliance for Genomics and Health

- Global Alliance for Genomics and Health has established 3 WGs including the Clinical Experts WG. They are now doing a mapping exercise to analyze the situation in the Cancer and RD communities to find priorities.
- A workshop is planned early March in UK, with an afternoon dedicated to discussion on Ontologies and Matchmaking (topics of the WG on Ontologies and WG on Genome/Phenome respectively).
- It is too early to know how Global Alliance will contribute as there is not yet any structure, funding etc., but it is essential to stay in contact with the Global Alliance and to highlight IRDiRC work.

Ontologies

Disease Ontologies

List of ontologies

- ORDO (Orphanet Rare Disease Ontology)
- ICD 9
- ICD 10
- ICD 11
- SNOMED

Recommendation

The WG agreed to propose ORDO as the disease ontology for IRDiRC to recommend.
This ontology was launched at the end of January (http://www.orphadata.org/cgi-bin/inc/ordo_orphanet.inc.php) in partnership with EBI. It integrates a nosology (classification of rare diseases), relationships (gene-disease relations, epidemiological data) and connections with other terminologies (MeSH, SNOMED CT, UMLS, MedDRA), databases (OMIM, UniProtKB, HGNC, ensembl, Reactome, IUPHAR, Geantlas) or classifications (ICD10). The mapping with HPO will be available in a few months.

Further information on the ontologies related to the WG members questions:

- A contact email is available to request changes, improvements and other mapping but a tracker could also be added.
- Terminology of the ontology is based on the most used terms in the literature to facilitate connection with other databases. For new syndromes, the policy is to use clinically relevant terms and to include synonyms that are searchable. The rules for nomenclature will be published on the Orphanet website.

Dissemination of the recommendation

- A webpage on Ontologies will be created on the IRDIRC website. Contents: definition of ontology, how to use ontologies, list of the disease/phenotype ontologies (with links to the webpage; not comprehensible but useful) and recommendations for the ontologies that should be used.
- Publication of a commentary (Nature Genetics, Am J of Human Genetics?)
- Publicity on several websites
- Announcement at events (RD-Connect meeting in February, etc.)

Phenotype Ontologies

List of ontologies

- HPO
- Orphanet
- Possum
- Snomed
- PhenoDB

Recommendation

The WG agreed to propose HPO as the phenotype ontology for IRDiRC to recommend.

International Consortium of Human Phenotype Terminology

This is a mapping instrument. A core phenotypic terminology of 2,300 terms was derived from all the terminology in use. Each term will have a definition and will be identified by unique identifier to be used in databases and mapped to other terminologies (HPO, PhenoDB, LOVD, Possum, SNOMED, Orphanet). The terminology needs to be reviewed and for some chapters to be completed by domain experts before publication on a dedicated website to be hosted by IRDiRC.

Publication of the terminology (within the next 4 months)

- If ICD and SNOMED are implicated as co-authors, it could be published in Nature Genetics – this should be the first target
- Another possibility would be to publish a series of commentaries in the Am J of Human Genetics: IRDiRC, Disease ontology, Phenotype ontologies, etc.
- In Europe: Eur J of Human Genetics, Orphanet J Rare Dis (free access is good for some countries).

Biobanks Ontologies

Biobanks ontologies may not exist yet. The only information that was found is an Ontology of administration of biobanks.

Possible resources:

- Elixir (European life sciences infrastructure for biological information) may be involved. This infrastructure aiming to share biological information is not specific to rare diseases. This would still necessitate people to provide the work. RD-Connect should be making the link with this infrastructure if necessary as this project includes specialists of biobanks and ontologies people).
- BBMRI: European infrastructure for Biobanking, funded by EC. However, it is unknown if ontologies is part of their project and their work is not specific to rare diseases.
- ORDR, which funds RD-Hubs, can be a source of information.

A challenge is to have biobanks repository to participate. Biobanks can register to be searchable. It is necessary to get more information.

Note from the Secretariat: the Interdisciplinary Scientific Committee has a WG on Biobanks.

Main deliverables

- Completion of the list of Ontologies to be sent to the Scientific Secretariat
- Preparation of the Ontologies webpage for the IRDiRC website.
- Contact the editorial board of Am J of Human Genetics to inquire if they would be interested in a series of commentaries about IRDiRC and its progress