IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 1st DSC WG Genome/Phenome teleconference

July 18, 2013

Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

Prof Anthony Brookes, Leicester, UK, chair
Dr Kym Boycott, Ottawa, Canada
Prof Han Brunner, Nijmegen, the Netherlands
Dr Xavier Estivill, Barcelona, Spain
Prof Ada Hamosh, Baltimore, USA
Prof Jim Lupski, Houston, USA
Prof Olaf Riess, Tuebingen, Germany

Ms Diana Désir-Parseille, IRDiRC Scientific Secretariat
Dr Sophie Höhn, IRDiRC Scientific Secretariat

Apologies

Dr Wei Wang, Shenzhen, China
Dr George Patrinos, Patras, Greece

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REPORT

General introduction

The Diagnostic Scientific Committee (DSC) is composed of 9 members for the moment and oversees 4 WGs:

- WG on Model Systems
- WG on Genome/Phenome
- WG on Ontologies and Disease Prioritization
- WG on Sequencing

Each WG will bring new insights and strategies to be considered by the DSC. The Chair of the DSC will report the priorities identified by the WG to the Executive Committee who will next meet in September 2013. This bottom-up approach will allow better efficiency and will keep the patient at the heart of IRDiRC missions.

WG tasks

Five major aspects of the WG on Genome/ Phenome were raised during the discussion:

- The WG will focus on near-term and achievable goals
- The principle goal of the WG is to enable the interpretation of genetic variation in RD
- The WG will seek to enable the maximal exploitation (via exposure, discovery, aggregation, and controlled and open sharing) of all genotypic and phenotypic data and knowledge
- The WG will advise on a suitable data ecosystem to facilitate progress in the field
- The WG will define what is necessary in the context of IRDiRC objectives and at policy level

IRDiRC connection with the Global Alliance initiative

Context

The WG discussed the Global Alliance initiative – a major new international initiative seeking to promote genetic data sharing. Its major goals concern establishing standard technologies and procedures to maximize the use made of genotype and phenotype data. A white paper was released early in June, describing the problem, the mission, the strategy, and options for stakeholder's involvement, to enable responsible sharing of genomic data. Within just a few months the Global Alliance had gathered >80 major organizations via a signed Memorandum of Understanding. IRDiRC has joined this initiative.

The mission of IRDiRC overlaps significantly with that of the Global Alliance, thereby providing it with a specific and distinctive expertise around clinical and research data about RD patients. All WG members agreed that regarding the specificity of RD and the shared goals, IRDiRC will seek to take the lead on specific tasks and activities in the RD area, to become the voice of RD in the Global

Alliance. The Genome/Phenome WG will help in framing and phrasing their outputs as a proposal for such a project.

Combining phenotype with genotype data is one major goal of the Global Alliance initiative, and the WG recognised that since RD patients are very engaged and committed, a bottom-up lead and drive should be emphasized.

Action plan

The WG is planning to write a proposal to the Global Alliance initiative that will need official adoption from the Executive committee.

This proposal will:

- Identify relevant areas where IRDiRC would be the natural leader
- Identify similar initiatives in different countries, their limits and solutions
- Influence future funding opportunities

The argumentation for such a proposal is:

- Distinctive and clinically actionable data from the RD field
- Engagement of patient community, in concert with researchers and clinicians
- An ecosystem for data discovery, aggregation, and sharing, with disease experts involved

The strategy would be to:

- Discuss with the Executive Committee the best format of this proposal
- Engage in discussion with Patients' organizations and members of the Executive Committee to bring strong support to this initiative
- Consult other WGs, especially those supporting the Interdisciplinary Scientific Committee

Ecosystem

Discussions about the data collection/connection ecosystem covered three major issues:

- How to address the challenge of pathogenicity inference?
- How to promote the discovery of new disease-causing genes?
- How to connect data from diagnostic and research laboratories and patient registries?

Opportunities and issues identified

- Overlaps of clinical and research databases
- Overlapping phenotypes for different RD
- Patient consent for genome/exome research
- Need of 'integration' databases/portals (e.g., national or disease focussed) in the combined federated and centralized data management ecosystem
- Existing disease-focused data resources

- Importance of integrating the data source (laboratory, clinics, patients) for complete understanding and representation of disease characteristics and natural history
- Difficulties of implementing compatible standards in large numbers of databases
- Lack of support from funders for tools and interfaces that will bring database interoperability
- Local and global storage needs and query systems

WG propositions

- Create national hubs and disease-focussed portals as the integration layer (and tools and standards for the creation of these) within the total architecture
- Financing the integration layer & its components through IRDiRC structure
- Identifying and supporting local hubs for 'back-to-source' queries
- Making suitable data, knowledge and aggregated information available (by direct sharing and/or API mediated discovery or access) for use by global systems
- Defining the range and representation of phenotype data for association with genomic data within the total ecosystem

Main deliverables

- Circulate the Global Alliance's white paper to the WG members
- Discuss the format of the proposition to Global Alliance with the chair of IRDiRC
- Send a doodle to plan the next teleconference to be held the last week of August