IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

Meeting report series

Report of the 1st DSC Working Group on Model Systems teleconference

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Organization

Organized by: IRDiRC Scientific Secretariat
Teleconference

Participants

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REPORT

Introduction of the Working Group

An overview of the Working Group on Model Systems was first presented. Members were reminded that emphasizing the importance of model organisms (yeast, worms, flies, zebrafish, mouse...) is necessary on an on-going basis. That awareness of the importance of basic science (understanding gene function and pathway mechanisms, and how function is important for developing rational approaches to management, prevention, and treatment of disease), and the importance of having a rational approach to disease, goes through cycles of people really appreciating it and less appreciating it. This affects funding, among other things. This WG is a great opportunity to stimulate many connections among basic scientists working in model organisms, medical geneticists, and clinician scientists and thus to develop new possibilities for genome projects. This WG will have an impact if it produces recommendations to the larger body, particularly in terms of funding. Following the introduction of the WG, all the participants briefly introduced themselves.

Overview of IRDiRC

IRDiRC was launched in 2011. It is governed by an Executive group of funders (now 34) that will make decisions about priorities for funding in rare diseases. The two main goals of IRDiRC are:

- Produce diagnostic tools for a majority of rare diseases by 2020
- Develop 200 new therapies for rare diseases by 2020

The Executive Committee decided that there would be three Scientific Committees:

- The Diagnostic Scientific Committee
- The Interdisciplinary Scientific Committee
- The Therapies Scientific Committee

Each of the funders has contributed 10 million USD or has designated 10 million USD of funding to go towards rare disease efforts in their country. When becoming a member, they can nominate one person to one of the Scientific Committee. The three Scientific Committee chairs wrote the policies and guidelines approved by the Executive Committee, and created twelve Working Groups (WG), four for each Scientific Committee.

The WG under the Diagnostic Scientific Committee are:

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

The Working Groups have to identify the gaps where international cooperation could make improvements, areas that should receive more funding, and coordinate current research. This is

reported by the Scientific Committee. The chairs of the Scientific Committees attend twice a year the Executive meetings. The next Executive meeting is scheduled in September in Miami (USA).

Role and goals of the WG on Model Systems

Catalyzing connections and collaborations

How could the WG get the information about the genetic variants causing human disease to researchers that have expertise and experimental capabilities to approach the functional consequences of specific mutations? Between 3 000/4 000 rare disease genes will be identified over the next 5 years, translating to several genes being identified per day. This is a huge amount of information for analysis, and a tremendous opportunity. In Canada, the Canadian Federal Funding Agency has established a Committee called "Models and Mechanisms to Therapies" which raises awareness that model systems are still very relevant to human diseases and is advocating funding mechanisms to allow rapid funding of functional studies in model system that will inform rare disease gene mutations being identified.

Initiatives that could be developed to facilitate collaborations:

- Establish a mechanism to establish connections and collaborations at the pre-publication time between the model organism groups and the clinician scientists
- Contact the laboratories that could functionalize rare disease mutations being discovered, and invite them to submit a one page proposal in order to connect them quickly
- Create an open model systems 'market place' for rare diseases with accessible information
    - Centralized platform with a call for collaboration
    - Useful for gene discovering and for gene modeling
    - Relevant for therapy and diagnosis
    - Standardization of information deposited would be required (clinical phenotype, variation data, functional information)
    - Quality control of the information would be necessary (for example, issue of partial phenotypic information)
    - Recruitment of experts: a communication strategy is required
    - Confidentiality would be required (competition issue)
    - People would have to agree to rules of regulation
- Help develop mechanisms for people mobility
    - Training networks from the UE
- Promote specific researchers and specific institutions/centers involved in rare diseases in order to integrate their knowledge

Identifying specific opportunities

- Places where there may be opportunities for collaboration
- Other areas with opportunities for growth

Gaps in resources that are needed

- Places where gaps in resources could be filled by IRDiRC infrastructures

Communication to the model organism community

- Contact the Model Organism meeting organizers (annual yeast, worm, fly, zebrafish and mouse meetings) to propose a special session to describe the updates and opportunities in rare disease gene identification and links to model systems.

Other issues and concerns

- Knock-out models and phenotypes
- Use of bioinformatics
- Create a model systems working map
- Try to know more about what is happening in IRDiRC countries
- Create an updated list of rare genetic disease
- Find phenotypes that model systems could predict in humans

Next steps

- Have specialized meetings to bring people together
- Publish an editorial piece or a journal article to be more visible

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 at the next teleconference
- Write a paragraph on the idea of a “market place” and information resource
- Write a paragraph on literature resources
- Write a paragraph on the idea of a short-term communication strategy with the model organism communities
- Send a doodle to plan the next teleconference to be held in three or four months
- Arrange a meeting of WG members and guests during the ASHG meeting in Boston