Moving Towards New Rare Disease Research Goals: IRDiRC Elaborates its Roadmap for 2018

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Introduction

IRDiRC, officially launched in 2011, was originally conceived with two main goals: to contribute to the development of 200 new therapies and the means to diagnose most rare diseases by the year 2020. Considerable progress on these goals has been made: the goal to deliver 200 new therapies was achieved in early 2017 – three years earlier than expected – and the goal for diagnostics is within reach. These accomplishments were celebrated at the 3rd IRDiRC Conference in Paris, France, in February 2017.

Capitalizing on the momentum of this progress, IRDiRC devised a new set of global rare disease goals for the decade 2017-2027. IRDiRC aims to accelerate progress with three goals for the Consortium, and ambitiously push the limits of what is currently possible in the longer term with an audacious vision for the field, all with rare disease patients’ lives in mind.

Delineation of New IRDiRC Goals

| Brainstorm of potential new goals |
| Discussion and refinement with Consortium Assembly and Scientific Committees |
| Presentation of preliminary goals and vision at the 3rd IRDiRC Conference |
| Integration of comments and further refinement |
| Vote on final goals and vision by Consortium Assembly |
| Publication of final goals and vision (peer-review articles and press release) |

Vision and Goals

**Vision:** Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.

In order to work towards this bold and ambitious vision, IRDiRC has set three goals for the next decade:

**Goal 1:** All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

**Goal 2:** 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

**Goal 3:** Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.

Next Steps: Roadmap 2018

To tackle these goals, following extensive and collective discussion and prioritization, the Consortium defined its Roadmap for 2018 which includes actions to accelerate research and development in rare diseases, and to remove numerous barriers and bottlenecks. These actions are aimed at improving research funding and coordination, identifying strategies to diagnose unsolved cases, facilitating data collection and sharing, advancing therapy development, boosting patient engagement in research and enabling international collaborations. IRDiRC Committees and Task Forces will deliver a series of recommendations for policies and standards, tools and infrastructure development, and adoption of best practices in response to the needs of the rare disease community.

Global Coordination of Research Funding Efforts

This initiative aims to globally coordinate research funding to maximize the impact and coverage of rare disease research on the field. Through the development of a real-time tracking database that will consolidate past, future and potentially upcoming funding opportunities, IRDiRC will empower its members to align funding actions, strategically determine areas of focus and to undertake joint funding initiatives.

**Handbook to Support Orphan Drug Development**

This project aims to create a simple guidebook for academic and industry drug developers that delineates and describes the available tools and initiatives specific for rare diseases and their optimal applications.