



IRDiRC HIGHLIGHTS

Pre-announcement of New Task Forces

The International Rare Diseases Research Consortium is glad to announce the upcoming opening of **5 new activities to continuously advance toward IRDiRC goals** and support the rare disease community in accelerating the time to diagnosis and the development of and access to therapies. **The calls for candidates will be published between Q4-2021 and Q1-2022.**

1. Big Bang Project – Drug Repurposing Guidebook

Introduction

Developers in the rare diseases field, and particularly developers that are engaging in repurposing approaches are very diverse, ranging from big pharma but increasingly and more often to clinicians, not for profit organisation including patients-led initiatives. Hence the whole Rare Disease Community and in particular the patients would benefit from a guide helping developers to navigate the regulatory & development tools and resources, efficiently and effectively repurposing medicines in new rare disease indications.

Objectives

The objective of the Drug Repurposing Guidebook is to help developers (of all kinds) navigating the rare disease landscape and identifying specific tools and practices of relevance for repurposing projects. The creation of the Development Guidebook will focus on repurposing approaches, following the same successful methodology used for the **Orphan Drug Development Guidebook**, i.e. explore incentives, regulatory tools, initiatives, development tools ('building blocks') that exists or are missing for drug repurposing.

2. PLUTO Project - Disregarded Rare Diseases

Introduction

As of today, less of 6% of rare diseases have approved treatments and most of the drug development efforts are actually concentrated on a limited number of diseases. Many technical and social reasons may account for this: lack of disease knowledge and academic interest, pathophysiological complexity of many diseases, cellular localization of the defective protein (for genetic diseases), diagnostic complexity, and - possibly above all – extreme rarity. Although the existence of a large group of "disregarded" rare diseases is unanimously acknowledged by the rare disease research and development community, no specific analysis has been conducted so far to confirm this scenario and trying to identify specific

commonalities amongst these diseases, with the potential secondary aims to identify removable roadblocks that may foster future research and development also for these diseases.

Objectives

The PLUTO project aims at using an integrated database search approach to (1) identify the rare diseases that are currently under-attended by academic research and industrial development alike, (2) find what characteristics they have in common, and -through this analysis- (3) to understand what are the roadblocks shrinking the chances of seeing effective treatments developed for these diseases in the near future. Based on the results of this analysis, the working group also aims at providing potential recommendations to funders and developers to overcome existing limitations and roadblocks for research and development for these “disregarded” diseases.

3. Primary Care

Introduction

Individuals living with rare diseases typically present first, and often recurrently, to their primary care providers (PCPs). These PCPs can also provide a medical home and coordinating centre that is central to the patients on their rare diseases journey. PCPs are community based, may provide care to multiple family members and have an awareness of local resources. Primary care may be provided by physicians, such as specialists in pediatrics, internal medicine, family practice, genetic counsellors, nurses, nurse practitioners, pharmacists, physical therapists, occupational therapists, respiratory therapists and others. PCPs are therefore central to all aspects of patients’ experiences during their rare diseases journey, be it in shortening the diagnostic odyssey, data sharing, access to approved and investigational therapies, care coordination and social outcomes.

Objectives

The objectives of the Primary Care Task Force are to deliver a state of play and identify the key challenges and opportunities to advance IRDiRC goals with a focus on primary care. Focuses therein are likely to include access to diagnosis and therapies, coordination of care, multidisciplinary team approach at the local level, increased awareness of rare disease initiatives, sharing access to data and bio-specimens, patient engagement and outreach to everyone.

4. Telehealth

Introduction

Telehealth, the delivery of health-related services, information and education virtually and remotely, has become one of the most efficient ways to expand access to healthcare and health services to all populations, and has been shown to be especially beneficial to fill the gap in care for those hard-to-reach populations that may reside in rural or underserved settings, or that may have limited access to transportation, or are medically fragile and may have impaired mobility or other complicated health issues that make traveling challenging to meet with rare disease experts. In addition to travel costs, there may be limited resources to support a multiple-day visit with several sub-specialists.

Objectives

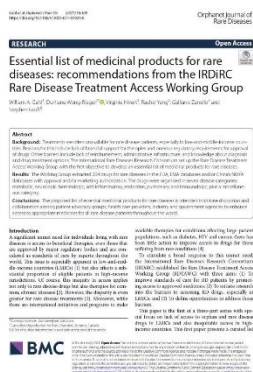
The Telehealth Task Force aims at (1) identifying the barriers to and opportunities for the use of telehealth to improve access to diagnosis, care, and research experiences for rare disease patients including, technological, legal, cultural,

linguistic, healthcare system, and patient/provider factors. (2) Survey existing models of telehealth to identify the factors that enhance or limit their adoptability and efficiency/ease of access in the rare disease community. (3) Develop “best practices” for introducing telehealth services into communities where they would be most beneficial using realistic and culturally-sensitive approaches, in partnership with local providers.

5. Rare Disease Treatment Access – Part 2

Following the completion of the first part of the **Rare Disease Treatment Access** working group and the publication of the first essential list of medicinal products for rare diseases (see below), IRDiRC will collaborate with **Rare Diseases International (RDI)**, the global alliance of patient-driven organisations, to develop the next phase of this activity. Through this collaboration, IRDiRC and RDI are looking to accelerate the identification of the barriers to accessing rare disease drugs into national markets, particularly in low-and-middle income populations.

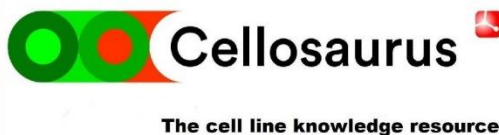
Publication of Essential List of Medicinal Products for Rare Diseases in Orphanet Journal of Rare Diseases



IRDiRC is pleased to announce that the recommendations from the **IRDiRC Rare Disease Treatment Access Working Group** have been published in the **Orphanet Journal of Rare Diseases**. The manuscript is titled ***“Essential List of Medicinal Products for Rare Diseases – Recommendations from the IRDiRC Rare Disease Treatment Access Working Group”*** and is authored by William A. Gahl, Durhane Wong-Rieger, Virginie Hivert, Rachel Yang, Galliano Zanello, and Stephen Groft.

[More Information](#)

New IRDiRC Recognized Resource: Cellosaurus



IRDiRC has recently accepted a new Recognized Resource, the **Cellosaurus**, a knowledge resource on cell lines aiming to describe all cell lines used in biomedical research. The Cellosaurus provides **information for about 130,000 cell lines and 25% of**

these cell lines are established from rare disease patients. For each cell line the Cellosaurus provides a wealth of information, cross-references and literature citations. The Cellosaurus is available on the ExPASy server (<https://web.expasy.org/cellosaurus/>) and can be downloaded in a variety of formats.

[More Information](#)

IRDiRC at National Press Foundation Workshop on Rare Diseases

IRDiRC members Lucia Monaco (Consortium Assembly Chair), Daria Julkowska, Durhane Wong-Rieger, Gareth Baynam, Samuel Agyei Wiafe, Anne Pariser, and Ritu Jain **discussed IRDiRC at a global scale** during a **two-day online workshop** entitled "Covering Rare Diseases" on **September 13th – 14th** and organised by the **National Press Foundation**.



[More Information](#)

Leadership and Membership Changes

- We congratulate **Marc Dooms** and **Birutė Tumienė** for their election as **Vice-Chairs** of the **Interdisciplinary Scientific Committee (ISC)** and the **Diagnostic Scientific Committee (DSC)**, respectively.
- We congratulate **Samuel Agyei Wiafe** for his election as **Vice-Chair** of the **Patient Advocates Constituent Committee (PACC)**.

IRDiRC IN EVENTS

IRDiRC held the following meetings:

- 9 July 2021: **Kickoff meeting** for the new task force on **Integrating New Technologies for the Diagnosis of Rare Diseases**. The co-chairs Sarah Bowdin and Clara Van Karnebeek were joined by experts from around the globe to discuss how best to meet our goals of identifying ways in which new technologies can aid diagnosis and to develop a clinical framework for their use.
- 25 August 2021: An in-person **strategic meeting** of the **Scientific Secretariat** was held in Paris to discuss the strategic direction of the functioning of the Consortium and the Scientific Secretariat in the coming years.



IRDiRC was presented at the following events:

- 7 July 2021: During a **United Nations High-Level Political Forum** side event titled "**Addressing the challenges of persons living with a rare disease as a human rights, sustainable development and equity priority**" by Dr. Lucia Monaco
- 8 July 2021: The **Clinical Research Network Task Force** was presented during a strategy meeting of the **European Joint Programme on Rare Diseases** by Dr. Lucia Monaco and Dr. Rima Nabbout

- 13-14 September 2021: During an online **National Press Foundation** workshop entitled “**Covering Rare Diseases**” (see details above)
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OTHER NEWS

National Organization for Rare Disorders (NORD) Rare Breakthrough Summit 2021

The National Organization for Rare Diseases (USA) is organising its annual NORD Summit 2021, **one of the largest multi-stakeholder events in rare diseases**, bringing together rare disease community stakeholders, including rare disease experts and leaders from patient advocacy groups, government, industry, and academia to discuss the most **current and critical topics related to rare diseases and orphan products**. The **fully virtual summit** will take place over two days from **October 18th – 19th**. For more information and to register, click on the link below.

[More Information](#)

EC publishes draft Strategic Research & Innovation Agenda (SRIA) for the Innovative Health Initiative (IHI)

The draft **Strategic Research & Innovation Agenda (SRIA)** for the **Innovative Health Initiative (IHI)** has been published by the European Commission (EC). The goal of IHI is to help create an **EU-wide health research and innovation ecosystem** that facilitates the translation of scientific knowledge into tangible innovations, with a **total proposed budget of €2.4 billion**.

The EC welcomes **feedback on the draft SRIA** from members of the rare disease community by email: **RTD-IHI@ec.europa.eu**.

[More Information](#)

Call for U.N. Resolution on Persons Living with a Rare Disease and their Families

[Rare Diseases International](#), [EURORDIS](#) and the [NGO Committee for Rare Diseases](#), along with other members of the global rare disease community and a Core Group of U.N Member States, have called for a **United Nations (U.N.) Resolution that recognises the complex challenges faced by persons living with a rare disease and promotes full participation and inclusion in society of all people**.

[More Information](#)
