

IRDIRC DECEMBER 2015 UPDATE for Executive Committee, Scientific Committees and Working Groups

Dr Christopher Austin elected as next IRDiRC chair

Dr Christopher Austin, Director of the National Center for Advancing Translational Sciences (NCATS) at the National Institutes of Health (NIH), USA, has been elected as Chair of the Executive Committee of the International Rare Diseases Research Consortium (IRDIRC). Dr Austin will follow in the footsteps of Professor Paul Lasko, Scientific Director of the Institute of Genetics of the Canadian Institutes for Health Research (CIHR).



When asked about his vision for IRDiRC, Dr Austin stated “Epochal advances in genomics; informatics, clinical medicine, and patient engagement over the last several decades have made rapid advances possible in the understanding and treatment of rare diseases. With that promise, the obligation has come to change substantially the performance of science and operations, to maximize the speed by which fundamental discoveries are translated into interventions that tangibly improve the health of people living with rare diseases. This requires a shift of focus from individual genes/diseases, researchers, and countries working in isolation, to a focus on connections among diseases and a research ecosystem that operates via rapid sharing of scientific insights, best practices, and projects, including a global community for rare disease clinical trials. These changes are particularly critical for rare diseases research given the number of rare diseases and the geographic dispersion of rare disease patients and researchers.”

“IRDIRC has made great strides since its founding in realizing such a global vision, and I would look forward as Chair of the Executive Committee to forging closer scientific and operational ties between founding members in North America and Europe, incorporating the efforts of (newly joined) members in Asia, Australia and the Middle East, and considering inclusion of countries with substantial rare disease burden but fewer resources,” Dr Austin added. “In addition, I would emphasize from a policy point of view not only the disproportionate burden of disease severity of rare disorders, but in this era of precision medicine the many lessons that rare disease research can teach about the diagnosis, translation, and treatment of newly segmented common disorders.”

Dr Austin, a neurologist and geneticist by training, has a lifelong passion for translational research in rare diseases that spans public and private sectors. Under his direction, NCATS researchers and collaborators

are developing new approaches and technologies to improve the efficiency and effectiveness of translational science, with the goal of getting more treatments and diagnostics to more patients more quickly, which coincides with the goals of IRDiRC.

IRDiRC wishes to thank Professor Lasko for his leadership and tireless outreach effort to bring name and recognition to IRDiRC and enlarging the consortium in recent years. Dr Austin's term will run from January 1, 2016 till December 31, 2018.

Upcoming IRDiRC workshops

- March 3, 2016 - Small-Population Clinical Trials Task Force - Workshop, London, UK

Upcoming IRDiRC teleconferences and meetings

- January 6, 2016 – Interdisciplinary Scientific Committee – Teleconference call
- January 11, 2016 – Executive Committee – Teleconference call
- January 14, 2015 – Operating Committee – Teleconference call (every second Thursday of the month, until March 2016)
- March 14, 2016, Morning – Individual Scientific Committee meetings – Lyon, France
- March 14, 2016, Afternoon – Joint Scientific and Executive Committees meeting – Lyon, France
- March 15, 2016 – Executive Committee meeting – Lyon, France

Rare disease research published on the website

- [Orphanet](#) becomes the 30th platform of the French Elixir Node
- [NORD](#) published the State Policy progress report: a state by state analysis
- The [European Commission](#) launched in public consultation a new notice on orphan medicinal products
- Public consultation on the EMA proposal to enhance early dialogue to facilitate accelerated assessment of priority medicines ([PRIME](#))

Research highlights from IRDiRC members

- [Genome Canada](#) Celebrates 15 Years of Research and Innovation
- The [NIH](#) will host its annual Rare Disease Day event on Feb. 29, 2016
- Very successful annual fundraising campaign for [AFM-Téléthon](#)
- [Lysogene](#) wins biotech award in Paris, France
- [NIH](#) unveils FY2016-2020 Strategic Plan
- New rare disease research collaboration between [PTC Therapeutics](#) and Massachusetts General Hospital
- Launch of the new EURORDIS' website for [International Rare Diseases Day 2016](#)

Research News

Interactive workshop on Patient-Centered Outcome Measures

On November 30, a workshop of IRDiRC's Task Force on [Patient-Centered Outcome Measures](#) (PCOM) took place in Paris, France. The purpose was to identify what should and can be done to support the development of patient-relevant outcome measures for rare diseases, in order to improve the feasibility and quality of forthcoming trials and to provide data of relevance to the patient community and other decision makers.

The workshop focussed on what is transferable to rare diseases of the experience gained so far with common diseases. The discussions in the morning addressed three aspects of the issue: how to develop PCOM, how to find existing tools to be adopted or adapted, and how to evaluate and expand on those tools. Several real-life examples with in different clinical trials were given; how well set-out PCOMs have contributed to successful clinical trials or the reverse. The next session was dedicated to ways to help and

support the rare disease community, including how to raise awareness about PCOM; the development of training tools; the different databases of outcome measures. The last session centered on the development of PCOM for RD; ways to share expertise; organization and funding of the development and guidelines for the development. The workshop ended with the identification of next steps and decisions on how to report on the workshop outcome.

The GA4GH-Framework, DECIPHER, ICHPT and HPO receive the “IRDIRC Recommended” label

On November 12, four new resources received the “IRDIRC Recommended” label, being [GA4GH's Framework for Responsible Sharing of Genomic and Health-Related Data](#), [DECIPHER](#), [Human Phenotype Ontology](#) (HPO), and the [International Consortium of Human Phenotype Terminologies](#) (ICHPT).

The *Framework for Responsible Sharing of Genomic and Health-Related Data* provides a principled and practical framework for the responsible sharing of genomic and health-related data. It contains foundational principles and core elements for responsible data sharing. The framework was praised for its general and robust governance framework and its clear methodology and involving a wide range of stakeholders.

The Wellcome Trust Sanger Institute's *DECIPHER* is a database and web-based platform enabling the deposition, analysis and sharing of phenotype-linked plausibly pathogenic variation in patients with rare genetic disorders. By making consented, linked- anonymized patient data available, DECIPHER facilitates collaboration and fosters communication between clinicians and scientists around the world. It was commended for its solid background, being embedded in a strong research environment. As such, DECIPHER was reviewed as an excellent resource to exchange clinical and genomics information on patients that suffer disorders for which the diagnosis is unknown.

HPO aims to provide a standardized vocabulary of phenotypic abnormalities encountered in human disease. The HPO is currently being developed using cross references to the medical literature, Orphanet, DECIPHER, and OMIM. HPO was positively evaluated as resource that provides the best existing system to standardize phenotypic information in rare diseases registries. An additional focus was put on the number of tools that are currently already developed to improve the use of this ontology provides the added value for the analysis of undiagnosed cases affected by unknown rare diseases.

ICHPT provides the community with a set of terms to describe phenotypic features to be used by any terminologies to achieve interoperability between databases, in particular to allow the linking of phenotype and genotype databases for rare diseases. ICHPT was positively assessed for its facilitation of standardization in the description of rare disease features and for gathering data efforts.

IRDIRC-related calls

AFM-Telethon has launched its [medical](#) and [scientific](#) calls. These calls aims to support research which will increase our understanding of the neuromuscular system and/ or encourages the development of therapies for neuromuscular diseases and rare genetic diseases.

[Fondazione Telethon](#) has launched a call for a number of research projects (either single or multicentre) in basic or clinical research aimed at finding therapies for genetic diseases.

The NIH has launched a call for [natural history of disorders identifiable by screening of newborns](#). This call encourages applications that propose to develop studies that will lead to a broad understanding of the natural history of disorder

All calls can be found on the [IRDIRC-related calls](#) page.



Other News

Help wanted!

DNA diagnostics is based on sharing data on genes, variants and phenotypes. Without sharing, DNA

diagnostics would not be possible. When we do not share, we do not offer optimal care to the patients and their families.

.....but there is another side to this; the gene variant databases we all use in daily DNA diagnostics depends on volunteers, guardians of one or more genes and database curators. The curator is responsible for checking incoming data (submissions), promotes the database and adds data from literature.

The reward? You may write highly cited database updates, receives many thank you's from users, becomes instantly famous, become known as THE expert for the gene and have a respected addition for your CV.

The Leiden databases need more hands to cope with the work involved. It will not take much of your time and the work is highly appreciated. Still not convinced? Please realize that with 7 billion people on the planet and having only 25,000 human genes the chance of becoming a curator is as rare as competing in the Olympics. So mail to: databases@JohanDenDunnen.nl

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