

IRDIRC JULY 2017 UPDATE

IRDIRC and the Human Variome Project (HVP) are collaborating on Recognized Resources



A collaborative relationship has recently been established between IRDiRC and [Human Variome Project \(HVP\)](#) to highlight additional resources that advance research and development. **IRDIRC Recognized Resources** was created to highlight key resources which, if used more broadly, would accelerate the pace of discoveries and translation into clinical services. In a similar spirit, **HVP Recommended System** was designed to encourage the adoption of HVP standards and guidelines, and to inform users of resources known to be in compliance with these standards thus promoting data sharing and interoperability.

The HVP is an international non-governmental organisation that works to ensure that all information on genetic variation and its effect on human health can be collected, curated, interpreted and shared freely and openly. Following assessment, a number of tools and guidelines included in the vast HVP project are now also IRDiRC Recognized Resources: (1) [Mutalyzer](#), a software supporting checks of sequence variant nomenclature, (2) [Human Genome Variation Society \(HGVS\) Nomenclature](#), a set of recommendations to describe sequence variant in a consistent and unambiguous manner, (3) [Leiden Open\(source\) Variant Database \(LOVD\)](#), that provides a flexible, freely available tool for gene-centered collection and display of DNA variations, and (4) the [Gene/Disease Specific Variant Database Quality Parameter guidelines](#).

For those interested, please use the newly released service by LOVD to see if your country is sharing data on genes. Type the international country code in front of ".LOVD.org" and check, e.g. nl.LOVD.org for the Netherlands, au.LOVD.org for Australia. The data retrieved are from the LOVD3/shared database.

For more information on IRDiRC Recognized Resources, please read [this page](#).

Special attention to Prof Ségolène Aymé

The European Society of Human Genetics ([ESHG](#)), at the 50th Anniversary of the ESHG Conference, presented the ESHG Education Award to Prof Ségolène Aymé, Founder of Orphanet and former Coordinator of IRDiRC's Scientific Secretariat. With this award, the Society acknowledged her excellent work as a human genetics educator. She has crucially contributed to the development of professional and public policies in the field of rare diseases. She also initiated numerous projects in the development of patient care and rare disease research and education in Europe and beyond. The ESGH also recognised her important contribution as Chair of the ESGH's Professional and Public Policies Committee.



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Spotlights on IRDiRC Member Organizations

- NIH and collaborators identified a mutation in the *MYMK* gene as [the genomic cause for Carey-Fineman-Ziter syndrome](#).
- Karen Aiach, CEO of Lysogene, has been elected - with 14 others women - as [figureheads of the French Tech](#) (representing french start-ups). *Note: article in French.*
- A international collaboration between the Généthon and the Royal Holloway showed that the [injection of micro-distrophine could restore muscular strength in dogs suffering from Duchenne myopathy](#). The work was financially supported by the AFM Téléthon.
- The Japan Agency for Medical Research and Development (AMED) published a paper on the [Initiative on Rare and Undiagnosed Diseases \(IRUD\)](#) aimed to shorten the diagnosis time for rare disease patients.
- Publication of the [2016 activity report and 2017 action plan of EURORDIS-Rare Diseases Europe](#).
- A team of authors, representing the Joint Action for Rare Diseases ([RD-Action](#)), identified the [mechanisms influencing sustainability, equity and resilience of health systems for rare diseases](#).

Rare Diseases Research Highlights

- [Gene editing shows promising results in mouse](#) to treat Huntington's disease.
- A group of researchers published the [state of play of incentives supporting the development of advanced therapy medicinal products for rare diseases in Europe](#).
- An article published in the *Orphanet Journal of Rare Diseases* [discusses the value of the orphan drug designation to pharmaceutical manufacturers](#).

IRDiRC-Related Funding Calls

The Canadian Institutes for Health Research has launched its recurrent funding opportunity entitled : [Project Grant Program](#). This announcement is designed to capture ideas with the greatest potential to advance health-related fundamental or applied knowledge, health research, health care, health systems, and/or health outcomes. It supports projects with a specific purpose and a defined endpoint. Letter of intent: **August 15, 2017**. Application deadline: September 15, 2017.

The National Institute of Neurological Disorders and Stroke has launched a funding opportunity announcement entitled: [Clinical Trial Readiness for Rare Neurological and Neuromuscular Diseases \(U01\)](#). This announcement is to support clinical studies that will fill gaps in the design of upcoming clinical trials in rare neurological or neuromuscular diseases by validating clinical outcome measures or biomarkers, or by characterizing cohorts of relevant patients. Application deadline: **August 17, 2017**.

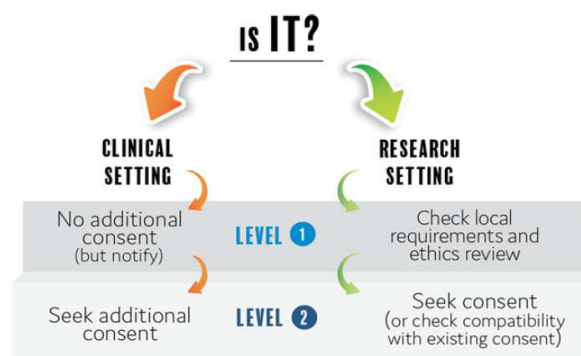
The French association [AFM - Telethon has launched a scientific calls for postdocs grants](#). This international call for proposals, open both to french and foreign groups, aims to support research which will: A- Increase our understanding of neuromuscular system and B- Encourage the development of therapies for neuromuscular diseases and rare genetic diseases. Application deadline: **September 5, 2017**.

For more IRDiRC-related calls, please consult [this page](#).

Featured Article

"Matching" consent to purpose: The example of Matchmaker Exchange

An article by Dyke *et al.* in Human Mutation discusses consent considerations and the resulting consent guidelines established by the Matchmaker Exchange (MME).



The Matchmaker Exchange (MME) was created to aid in the discovery of the causes of rare diseases through sharing of data from exome and genome sequencing performed in research and clinical care. The vast majority of patients still lack a clear diagnosis after initial analysis, but finding additional patients from across the world with a mutation on the same gene and exhibiting similar clinical manifestations can provide sufficient evidence to establish a diagnosis. To connect previously isolated effort, the MME was launched in 2013 to facilitate the identification of new disease genes through the international sharing of patients' genetic and clinical data.

One of the major issues confronted by the MME was to understand the consent requirements for such data sharing. A number of aspects were challenging to establish guidelines for patients' consent.

1- The projects using MME are generally **large data sharing initiatives that span across countries with different regulatory requirements**. For instance, the matchmakers currently connected through MME are located in four countries: [GeneMatcher](#), [myGene2](#) and [matchbox](#) are all three located in US, [PhenomeCentral](#) in Canada, [Patient Archive](#) in Australia and [DECIPHER](#) in the UK.

2- MME bridges **both clinical care and research**. One of the main differences between these two spheres is that while consent for clinical care is usually implied in the act of seeking care, research consent typically relies on written consent materials as a demonstration of proper awareness and agreement to a specific project.

3- MME data exchange occurs at two distinct data levels: Level 1 for matchmaking based on sharing candidate genes and clinical features, and Level 2 for matchmaking based on more detailed phenotypic information and/or DNA or protein sequence information including genomic variant datasets. These **two data levels present different amounts of privacy risk for individuals**, which also inform MME consent policy.

As MME bridges clinical care and research, the need for consent to share data in the MME depends on the context and objectives of the matching and on the potential harm introduced by sharing re-identifying information for matchmaking (see figure). Members of the MME drew up guidance on the content of consents materials for services participating in MME (based on Global Alliance for [Genomics and Health Consent Policy](#)).

For more details, read the [full article](#).

Upcoming Teleconferences and Meetings

- August 2, 2017 - Operating Committee - Teleconference
- September 18, 2017 - Interdisciplinary Scientific Committee - Teleconference
- September 25, 2017 - Solving the Unsolved Task Force - Teleconference
- September 28, 2017 - Funders Constituent Committee - Teleconference
- October 12, 2017 - Diagnostic Scientific Committee - Teleconference
- November 10-11, 2017 - Consortium Assembly - Face-to-face meeting in Tokyo, Japan

IRDiRC Documents

- Therapies Scientific Committee - [Report](#) of the 5th teleconference on 20 April 2017
 - Therapies Scientific Committee - [Report](#) of the 6th teleconference on 22 May 2017
 - Interdisciplinary Scientific Committee - [Report](#) of the 17th meeting - teleconference on 22 May 2017
 - Interdisciplinary Scientific Committee - [Report](#) of the 18th meeting - teleconference on 26 June 2017
 - Consortium Assembly - [Executive Summary](#) of the 4th meeting - teleconference on 12 June 2017
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Other news

We wish you all relaxing and sunny holidays!



If you are presenting in a meeting or a conference, and you would like to show some information about IRDiRC, standard slides are available on the IRDiRC private website; additional slides can be made available upon request.

Please also email the Scientific Secretariat when and where you will be presenting, so we can keep track of "IRDiRC presence at conferences."

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For more information on IRDiRC and its activities, please visit the [IRDiRC website](#). Stay up to date with news regarding IRDiRC and the rare diseases research community by following [@IRDiRC](#) on Twitter.