

IRDIRC OCTOBER 2014 UPDATE

for Executive Committee, Scientific Committees, and Working Groups



[IRDIRC Conference](#) program has been finalized.

New member of IRDiRC

- The [Saudi Human Genome Project](#) (SHGP) has officially been accepted as a new member of IRDiRC. Dr Sultan AlSedairy will be its corresponding representative. SHGP is a 5 year project which aims to build infrastructure and capacity in the rapidly advancing field of genomics in order to sequence ~100,000 Saudi genomes. This project will involve the creation of a national network of Genome Centers to recruit subjects and perform the sequencing work, as well as a centralized KACST Knowledgebase to store the resulting information on disease genes and make this available to enable future diagnostic screening efforts.



IRDIRC new documents

- Working Group on Population Controls Variant Datasets: [report](#) of the teleconference held on September 16, 2014

- Working Group on Sequencing: [report](#) of the teleconference held on September 17, 2014

Upcoming IRDiRC meetings and teleconferences

- October 31, 2014: teleconference of the Diagnostics Scientific Committee
- November 6, 2014: Executive Committee meeting in Shenzhen, China

News published on the website

- [The Children's Pharmacy Collaborative™](#): a tool to support pediatric drug repurposing
- [Patient-reported outcomes](#) should be included in clinical trials for rare diseases
- [Article series](#) on innovative approaches to study and treat rare diseases
- [Commission Call for Expressions of Interest for the position of member of the Committee for Orphan Medicinal Products \(COMP\)](#) as representing patient organizations
- The European Commission publishes its [implementation report on rare diseases: Europe's challenges and council recommendations](#)
- [Mitigating adverse effects of orphan drugs](#) despite challenges to conduct clinical trials on rare diseases
- Building [European Reference Networks for rare diseases](#) will require ongoing efforts to coordinate resources and data
- [Report from the international workshop](#) on biomarkers in Duchenne muscular dystrophy

IRDiRC-related calls

- The French Muscular Dystrophy Association (AFM, France) has launched the [Collagen VI Alliance call for projects 2015](#) to fund research into collagen VI deficiencies.
- The U.S. Food and Drug Administration (FDA) has launched a [Research project grant on clinical studies of safety and effectiveness of orphan products](#).
- The National Institutes of Health (NIH, USA) has launched the [Model Organisms Screening Center for the Undiagnosed Diseases Network](#)

(UDN) funding opportunity, which aims to establish a Model Organisms Screening Center for evaluating the pathogenicity and function of approximately 200 gene variants per year identified through the UDN.

Website updates

- The IRDiRC website has been reorganized to make it more accessible and user-friendly.
- The IRDiRC governance structure diagram has been updated with the addition of the Working Group on Population Controls Variant Datasets.
- The Become a Member section has been updated with the addition of the benefits of the consortium membership.
- In order to give more visibility to the clinical research funded by IRDiRC members, the information regarding research projects has now been divided into research projects and clinical trials.
- The IRDiRC presence at conferences section has been updated with the addition of one presentation about IRDiRC during the International Conference 'Rare Diseases: Europe's Challenges' that will take place in Rome, Italy on October 31, 2014.
- The IRDiRC in the news section has been updated with the addition of three article mentioning IRDiRC from Genome Medicine, Genetics and Expert Opinion on Orphan Drugs.

New list of research projects funded by IRDiRC members

- The Western Australian Department of Health list of research projects is now available from 2010 to 2013.



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