

NEWS



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



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IRDiRC Newsletter 9. September - October 2019



RE(Act) congress - IRDiRC conference 2020

[EARLY REGISTRATION](#) IS OPEN, [SUBMIT YOUR ABSTRACT!](#)

List of **speakers** is available [here](#).

11 - 14 March 2020. Berlin, Germany

The [BLACKSWAN Foundation](#) and [IRDiRC](#), the International Rare Diseases Research Consortium, are glad to announce the RE(Act) Congress and IRDiRC Conference 2020. We look forward to welcome scientists, researchers, patient organizations, and patients in Berlin.

[Read more](#)

LATEST IRDiRC NEWS

INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

State of Play of Research in the Field of Rare Diseases: 2015-2018

About IRDiRC State of Play of Research
The IRDiRC's report "State of Play of Research in the Field of Rare Diseases: 2015-2018" is a compilation of information published in scientific journals and press releases over the period of September 2015 to June 2018.

State of Play of Research 2015-2018
The report aims to inform stakeholders at large of developments in the field of rare diseases and support decisions of policy makers and research funders, as well as to inform the rare diseases community of the achievements and observed trends which shape the future of research and development for rare diseases.

<http://www.irdirc.org/irdirc-state-of-play-of-research-report/>

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LATEST IRDiRC NEWS



About IRDiRC Recognized Resources
IRDiRC Recognized Resources is a public label and quality indicator to designate resources (platforms, tools, standards, and guidelines) of fundamental importance to the international rare disease research community.

MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome
Bibi Högl, 1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37,38,39,40,41,42,43,44,45,46,47,48,49,50,51,52,53,54,55,56,57,58,59,60,61,62,63,64,65,66,67,68,69,70,71,72,73,74,75,76,77,78,79,80,81,82,83,84,85,86,87,88,89,90,91,92,93,94,95,96,97,98,99,100,101,102,103,104,105,106,107,108,109,110,111,112,113,114,115,116,117,118,119,120,121,122,123,124,125,126,127,128,129,130,131,132,133,134,135,136,137,138,139,140,141,142,143,144,145,146,147,148,149,150,151,152,153,154,155,156,157,158,159,160,161,162,163,164,165,166,167,168,169,170,171,172,173,174,175,176,177,178,179,180,181,182,183,184,185,186,187,188,189,190,191,192,193,194,195,196,197,198,199,200,201,202,203,204,205,206,207,208,209,210,211,212,213,214,215,216,217,218,219,220,221,222,223,224,225,226,227,228,229,230,231,232,233,234,235,236,237,238,239,240,241,242,243,244,245,246,247,248,249,250,251,252,253,254,255,256,257,258,259,260,261,262,263,264,265,266,267,268,269,270,271,272,273,274,275,276,277,278,279,280,281,282,283,284,285,286,287,288,289,290,291,292,293,294,295,296,297,298,299,300,301,302,303,304,305,306,307,308,309,310,311,312,313,314,315,316,317,318,319,320,321,322,323,324,325,326,327,328,329,330,331,332,333,334,335,336,337,338,339,340,341,342,343,344,345,346,347,348,349,350,351,352,353,354,355,356,357,358,359,360,361,362,363,364,365,366,367,368,369,370,371,372,373,374,375,376,377,378,379,380,381,382,383,384,385,386,387,388,389,390,391,392,393,394,395,396,397,398,399,400,401,402,403,404,405,406,407,408,409,410,411,412,413,414,415,416,417,418,419,420,421,422,423,424,425,426,427,428,429,430,431,432,433,434,435,436,437,438,439,440,441,442,443,444,445,446,447,448,449,450,451,452,453,454,455,456,457,458,459,460,461,462,463,464,465,466,467,468,469,470,471,472,473,474,475,476,477,478,479,480,481,482,483,484,485,486,487,488,489,490,491,492,493,494,495,496,497,498,499,500,501,502,503,504,505,506,507,508,509,510,511,512,513,514,515,516,517,518,519,520,521,522,523,524,525,526,527,528,529,530,531,532,533,534,535,536,537,538,539,540,541,542,543,544,545,546,547,548,549,550,551,552,553,554,555,556,557,558,559,560,561,562,563,564,565,566,567,568,569,570,571,572,573,574,575,576,577,578,579,580,581,582,583,584,585,586,587,588,589,590,591,592,593,594,595,596,597,598,599,600,601,602,603,604,605,606,607,608,609,610,611,612,613,614,615,616,617,618,619,620,621,622,623,624,625,626,627,628,629,630,631,632,633,634,635,636,637,638,639,640,641,642,643,644,645,646,647,648,649,650,651,652,653,654,655,656,657,658,659,660,661,662,663,664,665,666,667,668,669,670,671,672,673,674,675,676,677,678,679,680,681,682,683,684,685,686,687,688,689,690,691,692,693,694,695,696,697,698,699,700,701,702,703,704,705,706,707,708,709,710,711,712,713,714,715,716,717,718,719,720,721,722,723,724,725,726,727,728,729,730,731,732,733,734,735,736,737,738,739,740,741,742,743,744,745,746,747,748,749,750,751,752,753,754,755,756,757,758,759,760,761,762,763,764,765,766,767,768,769,770,771,772,773,774,775,776,777,778,779,780,781,782,783,784,785,786,787,788,789,790,791,792,793,794,795,796,797,798,799,800,801,802,803,804,805,806,807,808,809,810,811,812,813,814,815,816,817,818,819,820,821,822,823,824,825,826,827,828,829,830,831,832,833,834,835,836,837,838,839,840,841,842,843,844,845,846,847,848,849,850,851,852,853,854,855,856,857,858,859,860,861,862,863,864,865,866,867,868,869,870,871,872,873,874,875,876,877,878,879,880,881,882,883,884,885,886,887,888,889,890,891,892,893,894,895,896,897,898,899,900,901,902,903,904,905,906,907,908,909,910,911,912,913,914,915,916,917,918,919,920,921,922,923,924,925,926,927,928,929,930,931,932,933,934,935,936,937,938,939,940,941,942,943,944,945,946,947,948,949,950,951,952,953,954,955,956,957,958,959,960,961,962,963,964,965,966,967,968,969,970,971,972,973,974,975,976,977,978,979,980,981,982,983,984,985,986,987,988,989,990,991,992,993,994,995,996,997,998,999,1000

About MARRVEL
MARRVEL (Model organism Aggregated Resources for Rare Variant Exploration) is an open access database and prioritization tool of rare human gene variants for study in model organisms.

<http://www.irdirc.org/7vmarrvel>

ARTICLE

IRDiRC's State of Play of research report

This IRDiRC's report is a compilation of information published in scientific journals and press releases over the period of *September 2015 to June 2018*. The report aims to inform stakeholders at large of developments in the field of rare diseases and support decisions of policy makers and research funders, as well as to inform the rare diseases community of the achievements and observed

New IRDiRC Recognized Resource: The Model Organism Aggregated Resources for Rare Variant Exploration (MARRVEL)

MARRVEL is a search engine that collects data from a number of human genomics and model organism genetics databases and displays it in a rapid user-friendly format. It was created in 2017 by a group of researchers in the Undiagnosed

trends which shape the future of research and development for rare diseases.

[Read more](#)

Disease Network at Baylor College of Medicine as a one-stop data collection tool to facilitate the use of public genetic resources to prioritize rare human gene variants for study in model organisms. MARRVEL is available [online](#) and is continuously updated.

[Read more](#)



IRDiRC New Publications

IRDiRC has three new [publications](#):

- The use or generation of biomedical data and existing medicines to discover and establish new treatments for patients with rare diseases - recommendations of the IRDiRC Data Mining and Repurposing Task Force. *Noel T. Southall et al.*, 2019 - [Task Force Data Mining and Repurposing](#)
- Model consent clauses for rare disease research. *Minh T. Nguyen et al.*, 2019 - [Task Force Model Consent Clauses for Rare Disease Research](#)
- Ethical, legal, and social issues (ELSI) in rare diseases: a landscape analysis from funders. Adam L. Hartman et al., 2019. Working Group on ELSI.

Conference Presentations on IRDiRC

Here are the [past and upcoming conferences and meetings on IRDiRC](#):

- **September 20-22, 2019** - The 8th China Rare Disease Summit - Shenzhen, China
- **October 19-20, 2019** - 2019 China Conference on Rare Diseases (CCRD2019) - Beijing, China
- **October 19-21, 2019** - APARDO Summit 2019 Regional Collaboration for Global Change - Taipei, Taiwan
- **October 28-30, 2019** - XX Convention Scientifica, Fondazione Telethon. Riva del Garda, Italy
- **November 11-13, 2019** - The XIV meeting of the International Conferences on Rare Diseases and Orphan Drugs (ICORD) - Tel Aviv, Israel
- **November 12-14, 2019** - World Orphan Drug Congress 2019 - Barcelona, Spain
- **November 21 - 22, 2019** - IRDiRC Consortium Assembly, Face-to-Face Meeting - Paris, France

IRDiRC new members

IRDiRC is pleased to announce three [new partners](#) in the Consortium:

- **The French National Institute of Health and Medical Research (Inserm)** - the leading European organization for health research, and the coordinating institution of the European Joint Programme on Rare Diseases ([EJP RD](#)). Dr **Catherine Nguyen**, the Director of research of the Thematic Institute Multi-Organisms "Genetics, Genomics and Bioinformatics" at **INSERM**, was nominated as representative to the IRDiRC Consortium Assembly and Funder Constituent Committee.
- **YposKesi** - the first French industrial pharmaceutical company dedicated to producing gene and cell therapy drugs for rare diseases. Prof **Jean-Pierre Gaspard**, Executive Chairman at **YposKesi**, was nominated as

representative to the IRDiRC Consortium Assembly and Company Constituent Committee.

- [Eloxx Pharmaceuticals, Inc.](#) - a clinical-stage biopharmaceutical company developing novel RNA-modulating drug candidates designed to treat rare and ultra-rare stop codon diseases. Dr **David Snow**, Chief Business Officer at [Eloxx Pharmaceuticals Inc.](#), was nominated as representative to the IRDiRC Consortium Assembly and Company Constituent Committee.

IRDiRC video



Dr. Annemieke Aartsma-Rus's video on IRDiRC

Dr. Aartsma-Rus is a professor in translational genetic at the Leids Universitair Medisch Centrum and a member of [IRDiRC Therapies Scientific Committee](#). In this [video](#) she tells us why IRDiRC is needed in the rare disease field and why it is important have different stakeholders collaborating.

CONTACT US

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