

Embargoed until 11am JST, April 3, 2016

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BRCA Exchange aggregates publicly accessible BRCA1 & BRCA2 variants

KYOTO, JAPAN — The BRCA Challenge, a Demonstration Project of the [Global Alliance for Genomics and Health](#) (GA4GH), will release the newest version of the [BRCA Exchange](#) web portal on April 3, 2016 in advance of the annual meeting of the International Congress of Human Genetics (ICHG) in Kyoto, Japan. BRCA Challenge is an international effort to advance understanding of breast, ovarian, and other cancers by pooling genomic and clinical data on the genes BRCA1 and BRCA2. The BRCA Exchange, the first product of the BRCA Challenge, is a publicly accessible web portal that provides a simple interface for patients, clinicians, and researchers to access curated expert interpretations and some supporting evidence for genetic variants identified in BRCA1 and BRCA2.

Since its initial beta launch in October 2015, the BRCA Exchange has supported clinical decision-making by allowing any web user to search all BRCA1 and BRCA2 genetic variants that have been classified by the international ENIGMA consortium and to easily access the background rationale for classification. “The BRCA Challenge project represents the most comprehensive effort to date to pool BRCA variants from multiple international databases as a vehicle to provide supporting evidence for BRCA variant classifications in the public domain,” said Amanda Spurdle, chair of the ENIGMA consortium steering committee and Head of the Molecular Cancer Epidemiology Laboratory, QIMR Berghofer Medical Research Institute, Brisbane, Australia.

The new release will include an additional tier that allows web users, after agreeing to a disclaimer and data use policy, to interact with data on genetic variants that have not yet been expert classified. This represents the first time that all publicly available BRCA1 and BRCA2 variant data contained in databases such as ClinVar, LOVD, ExAC, and the 1000 Genomes Project, have been pooled in a single, federated resource. Such federation allows researchers, clinicians, and others to simultaneously search across the world’s collective knowledge on the genes.

“We are pleased that after such a short period, we have already received feedback from users around the world who have accessed the portal for understanding BRCA1 and BRCA2 variants,” said Stephen Chanock, Director of the National Cancer Institute’s Division of Cancer Epidemiology and Genetics, and co-chair of the BRCA Challenge’s Steering Committee. “We hope that the new release will add to its functionality and, eventually, to improve human health by offering better tools for understanding BRCA1 and BRCA2 genetic variation.” Notably, the updated portal integrates information on more than 13 thousand genetic variants from several national databases.

The BRCA Exchange will enable the development of a federated network to share BRCA data using the application programming interface (API) developed by the GA4GH Data Working Group. Such technical capabilities, developed in conjunction with members of the BRCA Challenge’s Evidence Gathering Group, will allow organizations around the globe to share their data directly with the BRCA Exchange. “By aggregating genomic and clinical data from around the world, the BRCA Exchange will significantly expand clinical impact because it will allow us to bring together currently siloed datasets to facilitate classifying variants of uncertain significance and resolving those with differing interpretations,” said Heidi Rehm,

Director of the Laboratory for Molecular Medicine at Partners Healthcare and member of the BRCA Challenge's Steering Committee.

Version 1.0 of the BRCA Exchange, which is expected to launch later this year, will offer a third tier of access, visible only to credentialed users. This space will allow for improved variant classification by aggregating and hosting case-level evidence.

"The BRCA Exchange will enable global engagement directly with data holders, clinicians, patients, and advocates," said Sir John Burn, Professor of Clinical Genetics at Newcastle University and co-chair of the BRCA Challenge's Steering Committee. "Aggregating global knowledge and making it freely accessible to the community will inform clinical care and improve expert classification efforts around the world."

Funded primarily by the US National Institutes of Health's BD2K program, the BRCA Exchange was built with significant expert input from the UC Santa Cruz Genomics Institute.

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The **Global Alliance for Genomics and Health** is an international, non-profit alliance formed to accelerate the potential of genomic medicine to advance human health. Bringing together over 350 leading organizations working in healthcare, research, disease and patient advocacy, life science, and information technology, GA4GH Members are working together to create a common framework of tools, methods, and harmonized approaches and supporting demonstration projects to enable the responsible, voluntary, and secure sharing of genomic and clinical data. Learn more at: <http://genomicsandhealth.org>.

The **BRCA Challenge** is a Demonstration Project of the Global Alliance for Genomics and Health that aims to advance understanding of the genetic basis of breast, ovarian, and other cancers by pooling data on BRCA genetic variants from around the world and bringing together information on sequence variation, phenotype, and scientific evidence. Improved understanding of genetic variation in these genes has the potential to improve patient diagnoses and disease prevention. The BRCA Challenge is co-led by an international team: Sir John Burn (Newcastle University, UK) and Stephen Chanock (National Cancer Institute, USA) are co-chairs of its Steering Committee and its sub-groups are co-led by Gunnar Rättsch (Memorial Sloan Kettering Cancer Center, USA), Antonis Antoniou (University of Cambridge, UK), Heidi Rehm (Harvard Medical School, USA), Johan den Dunnen (Leiden University Medical Center, Netherlands), Amanda Spurdle (QIMR Berghofer Medical Research Institute, Australia), Fergus Couch (Mayo Clinic, USA), Robert Cook-Deegan (Duke University, USA), Kazuto Kato (Osaka University, Japan), and Baroness Delyth Morgan (Breast Cancer Now, UK).