

BRCA Exchange Aggregates Data to Inform Understanding of Cancer Risk

Database provides information on gene variants to clinicians, researchers and patient advocates around the world.

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A global resource that includes data on thousands of inherited variants in the BRCA1 and BRCA2 genes is available to the public. The BRCA Exchange was created through the BRCA Challenge, a long-term demonstration project initiated by the Global Alliance for Genomics and Health (GA4GH) to enhance sharing of BRCA1 and BRCA2 data. The resource, available through a website and a new [smartphone app](#), allows clinicians to review expert classifications of variants in these major cancer predisposition genes as part of their individual assessment of complex questions related to cancer prevention, screening, and intervention for high-risk patients.

The five-year BRCA Challenge project was funded in part by the National Cancer Institute (NCI), part of the National Institutes of Health, and through the [Cancer Moonshot](#). A paper detailing the development of the BRCA Exchange [was published](#) January 8, 2019, in PLOS Genetics.

“This project has yielded a meta-analysis of BRCA1 and BRCA2 variants collected from multiple sources to understand how experts annotate specific mutations in the two genes,” said Stephen J. Chanock, MD, director of NCI’s Division of Cancer Epidemiology and Genetics and lead author of the paper. “There’s an urgent need for sharing data in cancer predisposition research. The BRCA Exchange is proof of principle that large-scale collaboration and data sharing can be achieved and can provide the latest and best quality information to enable clinicians and individuals to improve care.”

Certain inherited variants in these genes can increase the risk of breast, ovarian, and other cancers by varying degrees, whereas others are not associated with disease. Clinicians and patients need to know whether a given variant is likely to be disease-associated (pathogenic) and how likely a pathogenic variant is to cause cancer (penetrance). Until now, the available data on the inherited variants in these genes were not aggregated in a comprehensive way.

The BRCA Exchange dataset is composed of information from existing clinical databases—the Breast Cancer Information Core, ClinVar, and the Leiden Open Variation Database—as well as population databases and data from clinicians, clinical laboratories, and researchers worldwide. It currently includes more than 20,000 unique BRCA1 and BRCA2 variants. More than 6,100 variants

in the database have been classified by an expert panel, the Evidence-based Network for the Interpretation of Germline Mutant Alleles, and approximately 3,700 of these variants are known to cause disease. The BRCA Exchange pools variants from data resources worldwide, which should lead to inclusion of rare variants that are very occasionally observed.

With a single-point-of-access website, the [BRCA Exchange](#) provides information on these gene variants to clinicians, researchers, data scientists, patients, and patient advocates. It also serves as a demonstration project showing that this kind of comprehensive data sharing—requiring collaboration across hundreds of organizations, the establishment of an infrastructure to house the information, and the development of data-sharing protocols—is possible for other cancer predisposition genes and, indeed, for genes associated with other diseases.

Next steps for the project include collaboration with additional global data generators and data holders, continued technical development, and increased engagement with patients and patient advocates around the world.

[This announcement](#) was originally published on the National Cancer Institute website on January 9, 2019.