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BRCA gene mutations raise risk of breast, ovarian and other cancers

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Co-Founded by Quest Diagnostics and Inserm, with LabCorp as First Member, BRCA Share is now the Largest Open BRCA Datashare of Curated BRCA Variants Clinically Suitable for Patient Management

BRCA Share, a public-private BRCA gene datashare initiative, today announced the public release of a large collection of new data on genetic variants in the BRCA1 and BRCA2 genes. Mutations of these genes raise the risk of breast, ovarian and other cancers. The findings are to be presented today at the 6th International Biennial Meeting of Human Variome Project Consortium (HVP6) held at UNESCO headquarters in Paris.

BRCA Share was co-founded by Quest Diagnostics and the French National Institute of Health and Medical Research (Inserm) in April 2015, with Laboratory Corporation of America® Holdings (LabCorp®) as the first commercial participant. The goal of the initiative is to share clinical, genetic, epidemiological and biological data on BRCA variants, particularly variants of uncertain significance, in order to improve the quality of laboratory diagnostics to better predict which individuals are at risk of developing hereditary breast and ovarian cancers, and to accelerate research on BRCA gene mutations. BRCA Share builds on a BRCA gene-data curation process developed by Inserm, Institut Curie, and Unicancer Genetic Group (UGG) with associated University Hospital Centers, using data developed over a decade of BRCA patient testing by the 16 UGG laboratories in France together with the same associated University Hospital Centers.

This new release adds variants from Quest Diagnostics and LabCorp, two clinical testing laboratory companies operating primarily in the United States, to the collection previously developed by Inserm through the Universal Mutation Database (UMD) project in France. The BRCA Share™ database now contains over 6,200 total BRCA variants, an increase of nearly 30% compared to the previous Inserm UMD database. Of these variants, 334 are newly identified pathogenic or likely pathogenic, increasing by about 20% the total number of pathogenic or likely pathogenic variants to 1,826.

The data include 375 BRCA gene variants whose role in cancer risk was previously uncertain. Of these, 93% are now classified as neutral or likely neutral, while the remaining 7% are now classified as pathogenic or likely pathogenic. Neutral gene variants are generally considered benign, or non-disease-causing, while pathogenic variants are gene mutations that increase an individual's cancer risk.

The findings are significant because they will contribute to well-informed patient management strategies. A patient with a pathogenic BRCA gene test result may consider options to reduce the risk of developing cancer in the future, including increased screening or prophylactic mastectomy or oophorectomy (removal of breasts or ovaries). Participating commercial laboratories notify physicians of changes in classifications for affected patients.

The BRCA Share Initiative is intended to help scientists, physicians and laboratory experts improve the interpretation of BRCA gene mutations for patient testing and cancer research. With the release of the new set of variants and clinical data, BRCA Share is the largest database providing access to high-quality BRCA genetic data that has been researched and curated to determine clinical significance. Other public BRCA data initiatives collect a mix of curated and uncurated BRCA variant information from participating laboratories. Data curation is an essential step in maximizing the likelihood that data is clinically appropriate and actionable. Variants that are curated have been individually researched and categorized to determine the likelihood that they confer increased cancer risk. Advances in scientific knowledge may lead to new variant discoveries and reclassifications.

"We created BRCA Share to accelerate BRCA science and bring clarity to BRCA patient testing, and in short order, that's exactly what this initiative has done," said Charles (Buck) Strom, M.D., Ph.D., FAAP, FACMG, HCLD, vice president, genetics and genomics, Quest Diagnostics. "This doesn't just benefit patients of Quest, or LabCorp, or Inserm's participating labs. This initiative benefits anyone in the global medical community seeking robust analysis of BRCA genetics based on shared pooling of clinical-grade data and expertise."

"The promise of BRCA Share was to significantly improve BRCA diagnostics and enhance patient care and treatment," said Marcia Eisenberg, Ph.D., chief scientific officer of LabCorp Diagnostics. "BRCA Share has already

delivered on that promise, and it will help physicians and patients make more informed monitoring and treatment decisions that can improve health and improve lives."

"BRCA Share demonstrates that public-private data sharing collaborations, funded by commercial parties, can hasten advances in medical research that will benefit patients. This first experience encourages Inserm Transfert to continue its development strategy on health databases whenever it is relevant with industrial partners," said Pascale Augé, Ph.D., CEO, Inserm Transfert.

"In little more than a year, this initiative has provided more than 1,300 new variants and has lifted the veil of uncertainty from 375 variants and identified 334 likely cancer risk variants," added Prof. Christophe Béroud, Pharm.D., Ph.D., leader of the "Genetics and Bioinformatics" research team, Inserm/Aix Marseille University (AMU) UMR_S910.

Research entities and individuals with a research-only focus on BRCA -- including physicians and patients -- can participate in BRCA Share at no charge. The new data is available, beginning today, at http://www.umd.be/BRCA1/ and http://www.umd.be/BRCA2/ . Commercial laboratories may participate in BRCA Share by paying an annual fee to Inserm determined on a sliding scale to fund research and administrative expenses.

Since the launch of BRCA Share, nearly 1,000 scientists from 49 countries have registered to access BRCA Share for research purposes.

To participate, BRCA Share commercial laboratory members must submit their BRCA variant data, which Inserm's variant team then researches and curates. When a variant is curated and/or reclassified, Inserm notifies BRCA Share members and uploads the new data into BRCA Share. BRCA Share's participating laboratories may then begin to use the new data to inform decisions about patient test results.

Members of BRCA Share are free to also share their BRCA data with other datashare initiatives.

Source:
http://www.questdiagnostics.com/
