

New report: Labs differ widely in BRCA testing protocols

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An international survey of genetic testing labs shows that - despite the availability of BRCA1 and BRCA2 testing for more than two decades - global protocols and standards are surprisingly inconsistent when it comes to analyzing cancer susceptibility genes and their many variations.

A multi-institutional team led by Amanda Toland, PhD, of The Ohio State University Comprehensive Cancer Center - Arthur G. James Cancer Hospital and Richard J. Solove Research Institute (OSUCCC - James) surveyed 86 genetic testing laboratories around the world to better understand their testing practices for BRCA1/2, known cancer susceptibility genes linked the types of breast and ovarian cancer passed down through families.

The vast majority of responding labs—93 percent—used modern next-generation sequencing technologies that allow for simultaneous screening of multiple genes in a single, advanced [test](#). Just six relied on Sanger sequencing methods, the traditional approach used for [genetic testing](#) prior to the availability of advanced genomic testing tools like [next-generation sequencing](#).

Overall, researchers found that laboratories differed widely in their approach to analyzing BRCA—including in the extent of variant confirmation, whether non-coding DNA regions were sequenced and the techniques used to detect large genomic rearrangements that could provide clues about future cancer risk.

"This is important because it means that patients could be getting a different level of accuracy in their genetic results, based on the level of testing beyond baseline BRCA1/2 testing - there are variants of these [cancer susceptibility genes](#) that could be missed by some approaches and which are important to know about in terms of overall [cancer](#) risk," says Toland. "Global best-practice guidelines for BRCA testing are needed to ensure consistency in testing for patients, regardless of where they obtain their testing."

Provided by The Ohio State University

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