What to do when gene test for breast cancer risk comes back uncertain?

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In 2013, U.S. actress Angelina Jolie learned that she had mutations in a gene called BRCA1, which conferred very high risks of breast and ovarian cancer. To obviate that risk, she opted to have a double mastectomy; in 2015, she had her ovaries removed, too.

Many women face similar choices on the basis of BRCA1 tests, which search the gene for mutations that are known to increase the risk of breast cancer. Geneticists have documented many such mutations, as well as those that are more benign. But BRCA1 is a big gene, and there's a lot about it that we don't know.

This means that a test might return with a "variant of unknown significance" or a VUS—mutations that, simply put, we know squat about. "Women who carry a VUS seek to make informed decisions about their bodies and their health," says Andrea Downing, an activist and BRCA1-mutation carrier. "It's very hard to do this without clarity about whether a VUS is harmful or benign."

To get that clarity, you could do painstaking experiments to work out what each of these variants does, but "clearly, no one's willing to do that because there are still 350 of them around," says Lea Starita from the University of Washington. Alternatively, geneticists could rely on computer programs that predict whether a variant is likely to be harmful or benign, which would be much faster but also terribly unreliable.

Read full, original post: Taking the Uncertainty Out of Genetic Screening for Cancer Risk