

23andMe discovers surprising genetic connections between breast size and breast cancer

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Using data from its unique online research platform, 23andMe, a leading personal genetics company, has identified seven single-nucleotide polymorphisms (SNPs) significantly associated with breast size, including three SNPs also correlated with breast cancer in a genome-wide association study (GWAS) now published online in *BMC Medical Genetics*. These findings make the first concrete genetic link between breast size and breast cancer risks.

These findings were made analyzing data from 16,175 female 23andMe customers of [European ancestry](#), comparing their answers to survey questions including bra cup size and bra band size to [genetic data](#) at millions of SNPs. The analysis controlled for age, genetic ancestry, breast surgeries, [breast feeding](#) status and pregnancy history.

"The 23andMe [research platform](#) is a robust source of new genetic discoveries and this study demonstrates that important scientific insights can come from the most unlikely places," stated 23andMe CEO and co-founder Anne Wojcicki. "Nearly 90 percent of our more than 150,000 customers participate in our online research allowing 23andMe to make discoveries faster and more cost effectively than traditional research models," added Wojcicki.

While some factors of breast morphology, such as density, are known to be directly implicated in breast cancer, the relationship between breast size and cancer is less clear. While breast size is heritable, this study is the first to identify genetic variants linked to differences in breast size.

"The findings in this study show that some of the same biological pathways underlie both normal breast growth and breast cancer," explained lead

author Nicholas Eriksson, Ph.D. "Some studies have found that larger breast size as a young woman is associated with a slightly higher risk for breast cancer. The genetic factors we found support this concept that breast size and breast cancer are related."

The novel loci associated with breast size are rs7816345 near ZNF703, rs4849887 and rs17625845 flanking INHBB, rs12173570 near ESR1, rs7089814 in ZNF365, rs12371778 near PTHLH, and rs62314947 near AREG. Two of these SNPs are strongly correlated with SNPs associated with breast cancer (those near ESR1 and PTHLH), and a third (ZNF365) is near another breast cancer SNP. The other three loci (ZNF703, INHBB, and AREG) have strong links to estrogen regulation and breast development.

"These results provide insight into the genetic factors underlying normal breast development and show that some of these factors are shared with breast cancer," concluded Eriksson. "While these results do not directly support the known epidemiological relationships between breast size and cancer, this study contributes to a better understanding of the subtle interactions between breast morphology and breast cancer risk."

These breast size/[breast cancer](#) results continue to show the power of 23andMe's methodology in combining self-reported data on a broad range of phenotypes gathered via web-based questionnaires with genotypic data derived from self-collected saliva samples.

23andMe had previously published its first proof-of-concept results in the journal *PLoS Genetics* in June 2010, which reported novel associations for unusual traits such as asparagus anosmia and photic sneeze reflex and replicated associations for

other common genetic traits, published the discovery of two novel genetic associations for Parkinson's disease in *PLoS Genetics*, and presented a major replication study of over 180 genetic associations in the journal *PLoS ONE*. In April of 2012, 23andMe published a study in the journal *PLoS ONE* which reported five novel significant genetic associations for hypothyroidism in the largest known genome-wide association study of hypothyroidism conducted to date. Most recently 23andMe published its finding of novel associations found for male pattern baldness, some of which are also linked to Parkinson's disease and prostate cancer, in *PLoS Genetics*.

Provided by 23andMe Inc.

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