Medical Xpress

DNA study reveals BRCA1 mutations in three sisters, prompts life-changing decisions

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Sisters left to right: Chris Swatfager, Cindy Larson, Carrie Patnode and Amy Scheid. Credit: Chris Swatfager

Sisters often share certain genetic traits, such as hair color and facial features. But for three sisters from Minnesota, they discovered a much deeper connection—a shared genetic mutation known to dramatically increase their risk for breast and ovarian cancer.

Their BRCA1 variants came to light through a Mayo Clinic Center for Individualized Medicine DNA sequencing study. The study highlights the importance of genetic screening and the potential for hereditary health concerns. The diagnoses have prompted each of the sisters to take preventative actions for themselves, and their families.

"Having this knowledge is lifesaving and life changing," says Chris Swatfager, the middle sister from Minnesota who was the first to undergo the DNA testing and receive the difficult diagnosis.

BRCA1 is an inherited condition that significantly increases the likelihood of developing breast and ovarian cancer during a person's lifetime. According to the National Institutes of Health, the breast cancer risk is 55%–72%, compared to approximately 13% for the general population. The risk for ovarian cancer ranges from 30%–60%, compared to approximately 1% for the general population. A parent with a BRCA1 mutation has a 50% chance of passing it down to their child.

Genetic screening reveals hidden health risks

Given Chris's medical history of breast lumps, and her sister Carrie's breast cancer battle 16 years earlier, she says a clear picture suddenly came into focus. However, her original motivation to participate in the DNA sequencing study was driven by the heartbreaking death of her youngest sister, Amy, who had a heart attack in 2020. It marked the second heart attack-related death in the family. Chris believed there must be a hereditary link to heart disease.

Chris had learned that the Mayo Clinic study was screening for three actionable hereditary conditions, including a high cholesterol disorder called familial hypercholesterolemia, which raises the risk for a heart attack.

The other conditions in the study were related to BRCA1, as well as BRCA2, which is associated with breast, ovarian, pancreatic, and prostate cancers. Additionally, Lynch syndrome was being assessed, which raises the risk of colorectal cancer.

Though none of these other conditions had crossed her mind.

To her surprise, Chris's results ruled out a genetic susceptibility to familial hypercholesterolemia, revealing instead a BRCA1 mutation.

After undergoing comprehensive genetic counseling and receiving guidance from her physician, Chris opted for a double mastectomy and hysterectomy to mitigate her cancer risks. She also shared her diagnosis with her sisters, who, in turn, pursued genetic testing and received their own BRCA1 diagnoses.

For Carrie Patnode, who had already battled breast cancer at the age of 48, the diagnosis has brought assurance that she can stay ahead of any new cancer developments with increased screenings and scans.

"I think I may have done things differently if the BRCA test would have been provided when I was first diagnosed with breast cancer," Carrie says. "Now, after everything I've been through, I figure I'm going in every six months and getting mammograms every year and doing MRIs every year, and I'm confident they're going to find anything that reappears."

Cindy Larson, the oldest among the sisters and a retired letter carrier, developed bladder cancer shortly after receiving her genetic test results. She has chosen to hold off on any preventative procedures related to her BRCA1 variant, guided by the advice of her Mayo Clinic physician, Sandhya Pruthi, M.D.

A new frontier in predicting disease risk

Dr. Pruthi specializes in breast cancer prevention, and she's working to pinpoint Cindy's genetic risk estimation related to her breast and ovarian cancer syndrome.

Dr. Pruthi says new and sophisticated genome sequencing technologies are opening the door to less invasive alternatives with the advancement of polygenic risk scores. The complex scores, compiled from a combination of data from thousands of a person's DNA variants, have the potential to offer precise insights into how patients should respond to a genetic mutation diagnosis associated with a cancer syndrome.

"Polygenic risk scores allow us to potentially determine a patient's 5-year, 10-year and lifetime cancer risk," Dr. Pruthi says. "It may help us determine if and when a patient should undergo preventative procedures now or opt for surveillance over the next decade."

In the meantime, Dr. Pruthi says the power of genetic testing lies in its ability to help identify patients at high risk for cancer, enabling earlier screening and mitigating the risk of metastatic cancer and premature deaths—something she sees all too often.

Quest to bring genetic screening into everyday health care

Mayo Clinic is advancing the idea of bringing genetic screening to routine patient care.

"We're also studying the health utilization needs of those patients affected. This knowledge will transform the delivery of care in the coming decade," says Konstantinos Lazaridis, M.D., the Carlson and

Nelson Endowed Executive Director for Mayo Clinic's Center for Individualized Medicine.

Dr. Lazaridis says the immense impact of population-scale genetic screening reaches far beyond the benefits to individual people. It is also about the biological family members of those affected.

"It provides a collective wisdom that can shape public health strategies, increase disease prevention, and advance individualized medicine. It empowers people to be proactive about their own health care and prevention." Dr. Lazaridis says.

For Chris, Carrie and Cindy, equally important to their own diagnoses is sharing the hereditary condition with their children and other blood relatives so they too can receive early disease detection.

"This affects generations of families and I hope genetic testing will soon become available for everyone," Chris says.

Provided by Mayo Clinic

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