| BY **ASHLEY M. BIGGERS**
| GRAPHIC BY **MIRELLE INGLEFIELD**

Genome Thyself

A recent Supreme Court ruling opens the door for more comprehensive and accessible genetic testing – and discovery of previously unknown links to cancer and other diseases.

FOR JAN COGGINS, a healthy, fit 50-something, back pain was the first domino. For this seemingly benign symptom, her primary-care physician prescribed physical therapy and rounds of tests, including a colonoscopy. Nearly a year after she first showed symptoms, Coggins received a diagnosis: ovarian cancer.

Although her story is typical of patients who develop this difficult-to-diagnose form of cancer – one often discovered only in its late stages – Coggins was an exception in two ways. She didn't have a family history of cancer, and she tested negative for alterations in BRCA 1 and 2, a gene whose mutations researchers have linked to breast and ovarian cancers. It wasn't until 2014 that Dr. Mike Janicek, medical director of gynecologic oncology at Arizona Oncology's Scottsdale Healthcare Medical Center, delivered a new finding: Coggins was positive for an alteration in the PALB2 gene, also linked to those cancers.

The finding, which enabled Janicek and Coggins to make informed decisions regarding her care and that of her family members, was only possible as genetic testing for cancer has become more accessible and affordable. In June 2013, the Supreme Court ruled human genes couldn't be patented, allowing companies other than Myriad Genetics – who previously claimed a patent on BRCA1 and 2, and thus controlled the testing process – to provide screening. The results have been greater affordability and access to genetic testing, an increase in screenings and better-informed preventive care and treatments plans.

"Overnight, we went from testing two genes to gene panels," Janicek says. The current panels evaluate 23 to 29 different genes and can identify many more inheritable causes of cancer.

Seventy-five percent of breast and ovarian cancers attributable to genetic mutations are connected to BRCA1 and 2. Mutations in other genes, including, in Coggins' case, PALB2, cause the other 25 percent, meaning until two years ago, these genetic ties were going undiagnosed. The broad panels can also detect 50 known hereditary syndromes that cause cancers, such as those of the prostate, pancreas, brain, thyroid and stomach.

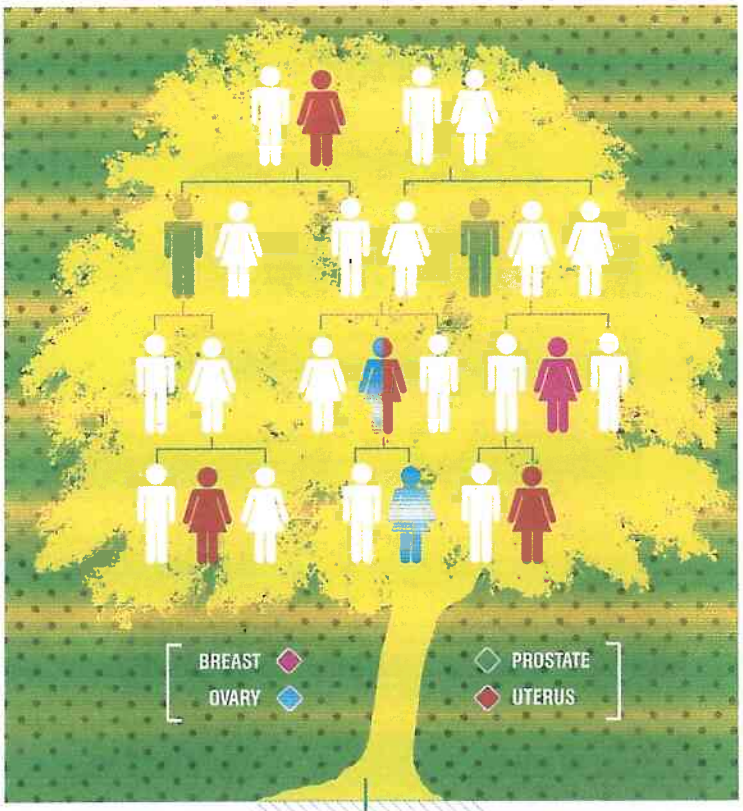
Of course, competition in the marketplace means lower prices – down from $5,000 to around $1,500 for a panel test. Patients can pay for genetic testing out of pocket without insurance authorization, but most insurance companies pay for the tests when a patient's medical history recommends it, e.g. when the patient has a family history of cancer, the patient's medical history includes cancer at an early age, or the history reveals an unusual cancer (such as male breast cancer).

Greater access and affordability has caused an uptick in people receiving genetic testing, and so has the "Angelina Jolie effect," created when the Hollywood star had a very public preventive mastectomy after receiving her results. Still, Janicek estimates practitioners are only diagnosing 10 percent of people with hereditary cancer syndromes, which he believes stems from lack of public awareness of and propensity for genetic testing.

Although the tests are 100 percent accurate, testing results are not perfectly predictive; patients may still develop cancer after a negative test, or live cancer-free lives with a positive one. For example, women with a BRCA 1 mutation have a lifetime risk of breast cancer of 46 to 63 percent; more studies are needed to determine the statistical risk for breast or ovarian cancer in patients with PALB2 mutations.

Navigating genetic test results is a delicate process, especially when the results have implications for family members, too. Dr. Janicek recommends working with one of the Valley's 21 board-certified genetic counselors to discuss the findings. Coggins' sisters both showed mutations in PALB2; as a result, one underwent a prophylactic hysterectomy.

Coggins' test results also helped her manage her medical care. Although she's already experienced the ovarian cancer her genetic test results indicate, PALB2 is also linked to breast and pancreatic cancer, for which she continues to undergo screenings. "I'm so glad I found this out so I won't be blindsided again by a cancer that I didn't know that I could contract," she says. "Information is powerful. This situation where you can know your genetic history is life-altering."



TREE OF GENETIC PREDISPOSITIONS

THE GRAPHIC ABOVE IS BASED ON AUTOSOMAL DNA CHARTS LIKE THE ONE PRESENTED TO OVARIAN CANCER SURVIVOR JAN COGGINS.

COGGINS HAS AN ALTERATION IN THE PALB2 GENE, WHICH INCREASES HER RISK FOR CANCER OF THE OVARIES, BREAST AND PANCREAS.

COGGINS' SISTERS ALSO BOTH SHOWED ALTERATIONS IN THEIR PALB2 GENES; AS A RESULT, ONE UNDERWENT A PROPHYLACTIC HYSTERECTOMY.

GENETIC TESTS CAN INDICATE GENETIC PREDISPOSITIONS FOR NUMEROUS OTHER CANCERS AND DISEASES, AS IMAGINATIVELY ILLUSTRATED ABOVE.