

Root & Branch

Genetic testing for cancer risk saved Sue Barbour's life—and the lives of at least 10 family members.

Thanks to the miracle of genetics, parents pass down all sorts of traits to their children. Some parents, for example, share their dimples. Some, blue eyes. Others bless their children with curly hair, freckles, left-handedness, cleft chins and widow's peaks. Children even inherit the ability, or inability, to roll their tongue.

In November 2018, however, Sue Barbour of Jackson, New Jersey, learned that she might have given something far more troubling to her children than a neat party trick or a unique hairline: multiple endocrine neoplasia type 2 (MEN2), a hereditary condition that's associated with an increased risk of thyroid cancer. The cause, a mutation of the RET gene that contributes to cellular growth and differentiation, is a loaded weapon that most carriers never realize is inside them. The fact that she found out she had it is a "miracle," according to Sue, who attributes her diagnosis to a series of serendipitous medical events that fell like dominoes in her favor.

"What I have is a very rare gene mutation. I was lucky that I found out I have it. My doctors said that almost never happens," explains Sue, 65, a paraprofessional who assists special-needs students in the sixth, seventh and eighth grades. "Everything that occurred—the sequence of events that brought me to where I am today—was very lucky. It saved my life."

Because she is a mother, grandmother, sister and aunt, it also saved the lives of at least 10 close relatives who have since tested positive for the same genetic mutation.

"It wasn't just me dealing with this," Sue says. "It was my whole family."



Sue Barbour, pictured here with daughter Louie Rivera and granddaughter Isabel Rivera, became the "chief medical coordinator" for her family, when a hereditary gene mutation associated with thyroid cancer was discovered.

Looking for Pancreatic Cancer

It all started in the most unlikely place: the bladder. Throughout the summer of 2018, Sue was feeling pressure in her urinary tract.

"Something just didn't feel right," recalls Sue, who initially assumed she had a urinary tract infection. When her symptoms persisted, she saw a urologist, who ordered a CT scan to check for tumors. "He said everything was fine, and then the symptoms just kind of went away."

Nevertheless, the urologist shared the CT scan with Sue's primary care physician, **Dina Meckael, D.O.**, who asked to see her. In the urologist's CT scan, it appeared as though a pair of cysts on her pancreas had grown. Her doctor therefore wanted her to see an oncologist.

"I was in shock, because my mother died from pancreatic cancer when she was 76. She found out she had it, and she died within a week," Sue says. "What I was hearing sounded like a death sentence to me."

Sue received a referral to surgical oncologist **Ronald Matteotti, M.D.**, medical director of the Hepatobiliary and Pancreatic Program and chief of Hepatobiliary and Pancreatic Surgery at **Jersey Shore University Medical Center**.

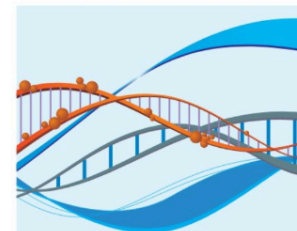
"I saw [Sue] in October 2018. She had an enlarged pancreatic cyst that was completely asymptomatic," recalls Dr. Matteotti, who ordered several tests, including a CTA scan and an endoscopic ultrasound. "Everything was completely within normal limits."

There were no indications of pancreatic cancer. But because of Sue's family history—not only her mother's pancreatic cancer, but also familial instances of breast, blood, prostate and colon cancers—Dr. Matteotti referred her to Nicole Salvatore, a cancer genetic counselor at Jersey Shore and **Ocean Medical Center**.

That decision set in motion a chain of events that permanently altered the trajectory of Sue's care.

RET Roulette

When Nicole saw Sue for the first time in November 2018, she commenced a game of genetic hide-and-seek. Specifically, she was looking for harmful mutations in a handful of genes that are associated with breast, ovarian, blood and pancreatic cancers. Instead of testing only for this



Genetics: A Crystal Ball for Cancer?

Gene mutations can be either inherited or acquired. When they're acquired, it's typically because of environmental factors, like smoking or radiation. When they're inherited, it's because one generation passes abnormal genes to the next.

The four most common types of hereditary cancer are breast, ovarian, uterine and colon cancers. If there are multiple people in your family that have had one of these cancers, and they were diagnosed under age 50, you should be referred for genetic testing.

Learn more at HMHforU.org/CrystalBall.

small group of genes, she conducted a large multigene panel test.

"It's like casting a wide net to see if there are mutations in any of more than 80 cancer genes that we know about," Nicole explains. "It's a good thing that we did that, because what came back positive was a mutation of the RET gene."

Suddenly, pancreatic cancer was no longer Sue's doctors' main concern. Instead, it was thyroid cancer, the risk of which is elevated as high as 90 percent in patients with RET gene mutations.

"This was a surprise to Sue because there's no history of thyroid cancer in her family," continues Nicole, who says it's possible her relatives had thyroid cancer but were never diagnosed with it because it was concurrent with other cancers.

Even more shocking to Sue than the results of her genetic testing was the suggested response to it: Because her particular gene mutation comes with an 87 percent risk of thyroid cancer, doctors recommended a thyroidectomy to remove her thyroid gland before it became cancerous.

"It was very overwhelming," says Sue, who was referred by Nicole to **Alexander Shifrin, M.D.**, surgical director of the Center for Thyroid,

Parathyroid and Adrenal Diseases at Jersey Shore.

"The medical guidelines are very clear: Every patient who is diagnosed as a carrier of [RET proto-oncogene mutation] should undergo a total thyroidectomy regardless of whether they have a tumor in their thyroid," Dr. Shifrin says. "There's no waiting and no observing. The thyroid needs to come out."

Dr. Shifrin, who performs approximately 350 to 400 outpatient thyroidectomies every year, removed Sue's thyroid on April 4, 2019, and discovered that a small amount of cancer had already taken root inside it, called medullary thyroid carcinoma.

"If I'd waited six months, it would have been too late," says Sue, who now takes a daily hormone replacement medication to compensate for her missing thyroid and receives regular follow-up care from **Danielle Lann, M.D.**, an endocrinologist dedicated to oncology at Jersey Shore.

Family Matters

What's most remarkable about Sue's story is that even as she was grappling with her own health care, she became "chief medical coordinator" for her family's health care.



Four of Sue's children, two brothers, one niece, one nephew and one grandchild have tested positive for the RET gene mutation.

"Because it's genetic, not only did I have to be tested [for the RET gene mutation], but my whole family had to be tested," says Sue, who has three brothers, five children, four grandchildren, and five nieces and nephews, all of whom were advised to undergo genetic testing. "My kids went right away and got tested, but my brothers needed a little more convincing to get the test. Initially they didn't want to know, but eventually they went and got tested."

Four of Sue's children, two of her brothers, one niece, one nephew and one of her grandchildren have tested positive for the RET gene mutation. So far, two of the group—one of her daughters and one of her brothers—have had their thyroids removed, and both were positive for cancer, like Sue. Other family members are also seeing Dr. Sullivan within the endocrine practice at Jersey Shore and plan to have the surgery soon.

"Sue has been the leader within the family as far as getting people in to be tested, even when they didn't want to get testing done," Nicole says. "She is a really unique patient, and she deserves a lot of credit for talking to her family members. She was willing to do whatever needed to be done to make sure they received the care they needed."

It wasn't easy for Sue. "I was a basket case," she says. "Even though I was born with it, I felt like it was my fault."

Fortunately, what she felt more strongly than guilt was gratitude. "If I didn't have the urinary problem that led me to Dr. Matteotti, I would have



Ronald Matteotti, M.D.

Board certified in surgery and integral and holistic medicine

800-822-8905

Neptune



Alexander Shifrin, M.D.

Board certified in surgery and endocrinology

800-822-8905

Neptune

never known any of this," Sue continues. "As weird as it sounds, I feel like my mother pushed me to go get that initial test, and all the tests after. Something made me do it."

It's impossible to say what made Sue go to the urologist in the first place. What gave her story a happy ending, however, is no mystery, according to her doctors: teamwork.

"The reason I came to Hackensack Meridian Health was because I'm a strong proponent of multidisciplinary care," says Dr. Matteotti, who practices alongside Dr. Shifrin and Nicole in HOPE Tower, where a state-of-the-art outpatient cancer center that provides integrative cancer care at Jersey Shore is located. "Especially when it comes to cancer treatment, we all work together to discuss patient cases and recommend the best treatment options, tailored to them."

Although he saw Sue for pancreatic cysts, not thyroid cancer, Dr. Matteotti was the catalyst that got Sue the care she ultimately needed. "Someone puts one and one together, then everyone else pulls on the same string," Dr. Matteotti continues. "If you just do isolated care, it's never to the benefit of the patient." ❄

GO ONLINE

Discover more about our **Hereditary Cancer Risk Program** that offers assessment for individuals who may have an increased risk of developing certain types of cancers at HMHforU.org/GeneticTesting.

John Dolan spent his life serving others, first as a firefighter in Orange, New Jersey, for 35 years, then moving on to his second love, building and fixing anything that needed attention. Whether it was building his daughter's house in Toms River 30 years ago, converting the porch of his home on the lagoon in Barnegat Bay into the ultimate outdoor man-cave, or being the handyman for anyone in the neighborhood in need, John loved serving others.

At 86 years old, after a long struggle with multiple progressive illnesses, including congestive heart failure and chronic obstructive pulmonary disease, John and his family sought information on home care services to support their own caregiving efforts to keep their husband and dad at home and as comfortable as possible.

"As my dad's condition declined, it became harder for my mom and our family to provide the level of daily care he needed," says John's daughter, Caroline Wettengel. "It was then that we found Hackensack Meridian Health At Home and Hospice and learned about home health care services and eventually hospice services."

"We felt good about our decision as soon as we started home care visits, which was almost eight months before my dad passed away in May. The caregivers knew exactly how to make my dad comfortable, whether it was something as small as adjusting his pillow to be just right, or bathing and dressing him in a way that provided him dignity and respect. These extraordinary caregivers took care of him as if he was their family."

Within a few months, John and his family were faced with a different conversation, one that included a topic

The Comforts of Home



Judith Dolan (center) with daughters Caroline Wettengel (left) and Mary Santambrogio (right). Judith's husband, John, received hospice care at Hackensack Meridian Health.

For one family, the fear of hospice is replaced with gratitude for a truly unique gift. By Larry Ginsberg



Theodore Ende, D.O.

Board certified in internal medicine

800-822-8905

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progressive conditions," he says.

"The hospice and care team at Hackensack Meridian Health brought joy to my dad's last months and became part of our family," Caroline says. "And their care for our family continued even after my dad passed away, with the care team attending my dad's memorial service and continuing to work with us during our period of mourning. I encourage families to learn more about hospice, as it is a gift of dignity, respect and love that is truly overwhelming." ❄

GO ONLINE

Do you have a life-limiting illness? Learn more about Hackensack Meridian Health's compassionate hospice care for peace and comfort in any setting at HMHforU.org/HospiceCare.

that is often unsettling and frightening: hospice care.

"Our experience with home health care for those months was so positive that when the time came to explore the idea, or even speak the word 'hospice,' it was less scary for all of us," Caroline says. "The Hackensack Meridian Health At Home and Hospice care team treated my dad as family, and it filled our hearts with joy that Dad looked forward to the care visits and they lifted his spirits. The hospice team literally took care of everything, which allowed us to focus and enjoy every moment with Dad."

John's primary care physician, **Theodore Ende, D.O.**, described the importance of including home care and hospice in care planning conversations. "Home health care provides so many benefits to patients and their families, from the actual clinical care from nurses, to personal needs from home health aides, to emotional support from social workers and others. I believe home health care and hospice are an important part of care planning and management for patients with chronic and serious