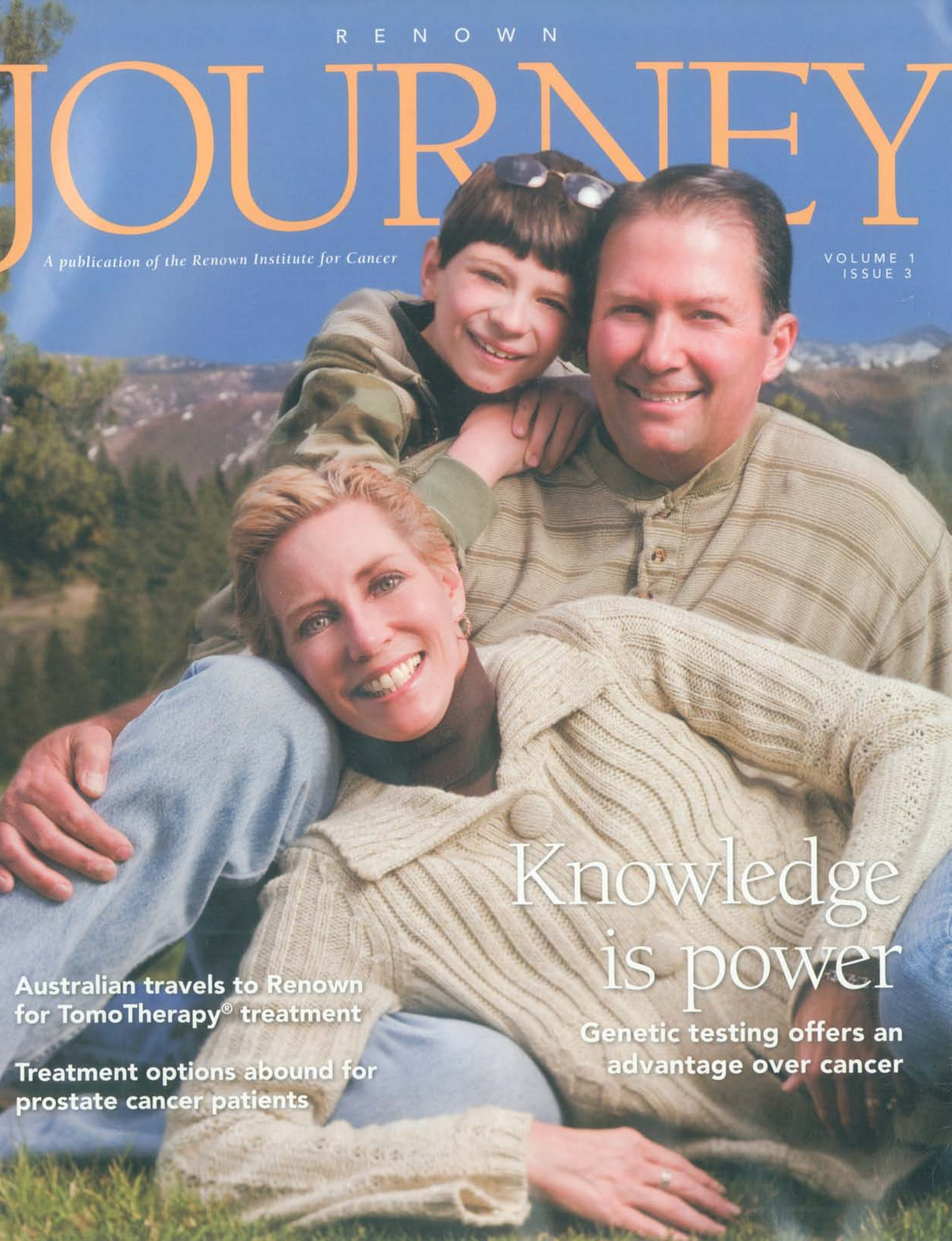
RENOWN

JOURNEY

A publication of the Renown Institute for Cancer

VOLUME 1
ISSUE 3



Knowledge is power

Genetic testing offers an advantage over cancer

Australian travels to Renown for TomoTherapy® treatment

Treatment options abound for prostate cancer patients

WRITTEN BY MIKALEE BYERMAN

Debora Amaral holds up a family photo. She and her relatives were tested for the BRCA2 mutation.

Knowledge is power

Three generations find strength and answers through genetic testing

THIS STORY HAS ALL THE COMPONENTS OF AN EDGE-OF-YOUR-SEAT MYSTERY NOVEL, the kind you read while wrapped in a blanket on a bone-chilling night: genetic mutations; sealed envelopes containing family secrets; an elaborate web connecting people across the country.

But for Truckee resident Debora Amaral, the story is reality. And as the mystery unfolded, her life came into clear perspective.

"I was first diagnosed with breast cancer in 2002," she said. "I went in for a routine mammogram with a girlfriend and my two-year-old baby in the waiting room. I came out thinking, 'Oh my God. I have cancer.'"

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Debora Amaral said genetic testing answered many of her questions, and impacted her healthcare decisions.

CANCER QUICK FACT:
Environmental (as opposed to hereditary) factors account for an estimated 75-80 percent of cancer cases and deaths in the U.S.

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The radiologist had almost instantaneously diagnosed a suspicious finding on Amaral's mammogram as cancer. But while the timing was unfortunate— her dad recently passed away, and she and her husband were building a home in Truckee — it wasn't entirely shocking.

"Cancer was well known in my family," she said, referring to her father's recent death from pancreatic cancer, her mother's bout with breast cancer almost two decades earlier and her sister's own breast cancer, diagnosed at the age of 35.

But Amaral considered herself lucky. Given her family history, doctors had recommended mammograms starting at an early age, thus precipitating her diagnosis of noninvasive cancer at the age of 42. A lumpectomy and six weeks of radiation later, she said she thought she was "good to go."

But just after celebrating her five-year-clear mammogram, she discovered a suspicious lump. This cancer was more invasive. And this cancer was the wake-up call her family had hoped would never come.

Amaral's oncologist noted that such a high incidence of cancer in the family was not normal, and was potentially the warning sign of an unusual form of cancer called genetic breast and ovarian cancer syndrome. These signs were noticed years ago, and Amaral's mother, Patricia Weeks, and sister, Suzanne, had considered genetic testing after seeing similarities in their medical histories — most notably Suzanne's cancer diagnosis at such a young age.

"Only 10 percent of breast cancers are hereditary," said Brandy Smolnik, MS, a genetic counselor who practices in Reno and counsels patients from the Institute for Cancer. "But Debora was younger than a typical age of onset. And coupled with her sister's breast cancer at 35, it was strongly recommended she see a genetic counselor."

Smolnik first sat down with Amaral and discussed her family history of cancer, ages of diagnoses and types of disease. Based on this information, a comprehensive diagram evolved, and Smolnik counseled her about how her family structure correlated to established risk models.

"We saw some evidence that this (genetic breast cancer) was a possibility in Debora's case," Smolnik said.

So the family — all three women, all breast cancer survivors — moved forward with the blood test.

"I only wish we had done this 17 years ago when we first started talking about this," Weeks said. "It might have saved Debora from some pain."

Such guilt, Smolnik notes, is common but unnecessary. "The reality is that so much is out of our control when it comes to genetics. We don't control what's passed on, and there are some wonderful genetic traits that we pass along as well."

Weeks did not test positive for the BRCA2 mutation, which is a genetic marker that indicates a predisposition for genetic breast and ovarian cancer. But both Amaral and her sister did.

"After all of that, the interesting thing was that I tested negative," Weeks said. "Their dad was the carrier."

With her newfound genetic knowledge, Amaral now knew how to proceed with her cancer treatment. Doctors were at minimum recommending a unilateral mastectomy (removal of one breast), but the outcome of the testing convinced her to choose the more radical but potentially life-saving double mastectomy. She also elected months later to have a complete hysterectomy (removal of the uterus) and an oopharectomy (removal of the ovaries) as a precaution to protect her from uterine and ovarian cancer.

> "I knew I had the gene. I knew the likelihood the cancer would return. And I knew I didn't want to go through this again."
>
> *Debora Amaral, cancer survivor*

"The testing answered so many questions, and I really looked at it as reassuring," she said. "I knew I had the gene. I knew the likelihood the cancer would return. And I knew I didn't want to go through this again. So I chose to clean myself out of everywhere this nasty cancer could hide."

And while Amaral recovered from her surgeries, another chapter of the story began to unfold: She now was concerned about her brother and his children, her sister's children and her father's sister, who has three children of her own.

"I turned it over to my mom," she said. "My choices were personal and based on my family's input, but we felt compelled to share the knowledge. It was empowering for us, but we also recognized its weight."

Smolnik says the choice to share genetic testing results can be a difficult one but ultimately gives the recipients of such news tremendous power.

"The benefits of learning about it (a potential genetic mutation) outweigh the negatives," she said. "If you have it, you have it — learning one way or the other doesn't make it suddenly appear. It just allows us to make better choices to deal with reality."

So for the daughters of Amaral's sister and brother, it will mean breast cancer screenings twice yearly. For the boys in the family, who are potentially at greater risk for melanoma and pancreatic cancer, it means greater vigilance in proactive screenings.

"I have a form letter that some patients send out to extended family," Smolnik said. She noted that the letter hints at some heavy news about the family's genetics, and it contains a sealed second envelope containing the details. "That way, it's their choice if they want to learn."

Weeks didn't have to go the route of the sealed envelope for her granddaughter, Suzanne's only daughter, Joy Lambert.

A 26-year-old news reporter and anchor in Alabama, Lambert said that watching her mother almost die when she was only 8 years old was "devastating."

"My brothers and I didn't understand," she said. "We watched her lie in bed, lose her hair — I'll never forget it. So obviously this is something I never want to go through myself."

She recalls learning that her mother had a tumor the size of a grapefruit in her breast before the cancer was even caught.

"The doctors discounted the tumor because she was too young," she said. "But the public needs to realize, no matter how young you are, it can happen."

Late last year, Lambert decided to be tested for the BRCA2 mutation — for which she tested positive. But like most of her family, she feels empowered by the knowledge, and it already has shown benefits.

"A month after my genetic testing, I had an MRI," she said. "They found a spot on my right breast. But it was biopsied and turned out to be a benign tumor. This shows that the doctors are taking this seriously, and I'm being proactive in my healthcare."

And she's also proactively telling the story, taking part in the countless forums available on bebrightpink.org, a Web site devoted to education and support for young women who are at high risk for breast and ovarian cancer. Additionally, she recently turned the cameras on herself, airing a news special called "Hope for Joy" that tracked her own story.

"Genetic testing is an incredible thing and saves lives," Lambert said. "It's scary to know, but it's scarier not to know. I'm positive I'll live a long life armed with this knowledge." ■

For more information about genetic breast cancer, visit www.renown.org. To access a Web-based tool allowing users to organize family health histories to share with their family doctor — or with other family members — visit www.familyhistory.hhs.gov. To follow up on Lambert's story, "Hope for Joy," visit www.renown.org/cancer.