completely. I was shocked,” said Joshalyn. “Halfway through my treatment course I went from receiving treatment once a week to twice a week and that is when things became extremely difficult. Unrelated to the trial, I developed a blood clot in my lung and carotid artery. Thanks to the close surveillance I was under while on the trial, Dr. Silber is the one that first noticed the fatigue I was experiencing was not normal, and decided to investigate further; that is when they found the clot.”

During the moments when Joshalyn was feeling like she wanted to quit, and she shared there were several, she would remember the women that came before her to even make this trial possible. She remembers seeing her mother go through treatment and continuing to work with little side effects, but this was a different cancer and a different treatment regimen. She was not able to continue working and faced financial hardship as a result. Following her treatment, Joshalyn underwent surgery to remove any remaining tumor, but no tumor was found. Her surrounding lymph nodes were also negative, meaning there was no invasive cancer present in the breast or lymph nodes; the trial had worked.

“Since I carry the BRCA1 mutation I decided to have a complete mastectomy and hysterectomy. Here I was, at the age of 34 with several body parts removed, no steady income, and all the plans I had for after my wedding put on hold. My advice is to take advantage of all the resources out there to help lessen the burden,” said Joshalyn. “And it may sound scary taking part in a clinical trial, but I would not be here today if it were not for the trial and the women before me that took part. I am proud to say that I am part of this story. I am part of this story. If you have to face cancer, you might as well try and help others in the process.”

After her experience, Joshalyn officially quit her job and used cancer as the motivation she needed to start her own business. Finally, she thought, ‘how hard could it be after fighting cancer?’ She had the opportunity to step back and think about what she was truly passionate about, and is now living her best life after starting a clothing company which has since grown to include handbags, shoes, and other accessories.

Now, almost five years out from her diagnosis, Joshalyn commented, “I am determined not to let this experience stop me, but only propel me forward in every aspect of my life. It was challenging facing this cancer at such a young age, mentally, physically, and financially. I share my story so that young women know it is possible, and that they should get to know their bodies and speak up when something isn’t right.”

Joshalyn experiences no lingering side effects, except that her nails continue to fall off. She encourages other women to consider clinical trials and will often speak to those considering a trial that are unsure. She is able to have an open and honest discussion about her experience, and offer advice and support.

I cannot say enough about the altruism and generosity I have seen from patients that take part in trials, it is truly unbelievable. Joshalyn faced every obstacle head on and took charge of her health. Her participation in this trial likely saved not only her life, but future lives as well.”

—Andrea Silber, MD

A NEW CHIEF AND NEW PLANS FOR CLINICAL CANCER GENETICS

Steve Kemper writer  Peter Baker photographer

Veda N. Giri, MD

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The field of genetic testing for cancer is rapidly evolving. Veda N. Giri, MD, the new Chief of the Division of Clinical Cancer Genetics at Smilow Cancer Hospital and Yale Cancer Center is at the evolution’s forefront. The field’s dynamism excites her, and she has ambitious plans for a novel, comprehensive program at Yale.

“T’m a medical oncologist by training,” Dr. Giri said, “and I’ve always had a deep interest in treating patients and in cancer risk assessment. Cancer genetics marries those two interests. Genetic testing has become central for informing us about better strategies for treating patients who carry specific genetic mutations, tailored screening, and hereditary cancer assessment for patients and their families.”

Dr. Giri joined Yale from Thomas Jefferson University, where she was director of Cancer Risk Assessment and Clinical Cancer Genetics since 2014. While the field of genetic testing has been grounded in assessing hereditary cancer risk, the field of precision medicine has skyrocketed the volumes of patients needing genetic testing to inform cancer treatment.

“Patients and providers are increasingly interested in thinking about what genetic testing should be done to inform decisions about cancer treatment,” said Dr. Giri. For example, multiple PARP inhibitors have been approved for use in the treatment of patients with a variety of cancers who carry mutations in DNA repair genes such as BRCA1 or BRCA2, due to clinical responses. As such, patients may be referred for genetic testing to inform cancer treatment, but it still requires that patients understand hereditary cancer risk, which can be uncovered by genetic results.

Genetic evaluation requires that patients receive information to make an informed decision for genetic testing. Since germline testing involves uncovering hereditary cancer risk, the first step, she continues, is explaining basic heredity to patients we inherit half of our DNA from each parent, and our genomes carry more than 25,000 genes. Genetic testing assesses genes of interest based on the cancer of concern and can also uncover additional cancer risks. For instance, a man with prostate cancer—one of Dr. Giri’s interests—may show a mutation in the BRCA2 gene, which signals risk not only for aggressive prostate cancer, but for pancreatic cancer, male breast cancer, and melanoma. Cancer risks for females with BRCA2 mutations include pancreatic cancer, female breast cancer, ovarian cancer, and melanoma. Since this is hereditary cancer testing, children have a fifty percent chance of inheriting mutations, and the associated cancer risks.

“There is a ripple effect in families, since the genetic mutation should be tested in all blood relatives and inform tailored cancer screening,” said Dr. Giri. One key reason for genetic testing, she adds, is to inform cancer screening and prevention. If that man with prostate cancer passes on a BRCA2 mutation to his son, Dr. Giri would recommend that the son start screening for prostate cancer at age 40 instead of age 50, which is in the recommendation for average-risk individuals. She also would discuss screening for pancreatic cancer especially if there is a family history of pancreatic cancer, and an annual clinical breast exam starting at age 35 for male breast cancer screening. If the patient’s daughter inherits the mutation, likewise, Dr. Giri would discuss screening for pancreatic cancer, and instead of an annual mammogram starting at age 40, Dr. Giri would discuss breast cancer screening starting at age 30 or younger based on family history of breast cancer with the addition of breast MRI. For females, the national guidelines also recommend risk-reducing surgery to remove the ovaries around ages 40-45 or after childbearing due to the higher risk for ovarian cancer in females who carry BRCA2 mutations.

“The testing can be lifesaving, as far as identifying hereditary cancer syndromes. It really has a population-level impact.”

Dr. Giri sees this intersection of genetic testing, clinical care, and population science as the future of cancer genetics. She has studied inequities in the availability of genetic testing for minority and underserved populations. To address the issue, she and her team developed projects to provide education on hereditary syndromes and the importance of genetic testing for individuals and their families.

One initiative recruited African American men and trained them as “peer genetic navigators” to educate individuals in their communities about prostate cancer and genetic testing. “The uptick of interest was remarkable,” said Dr. Giri. When the rising volumes of patients in need of genetic testing led to a relative shortage of genetic counselors, Dr. Giri devised an inventive work-around: an educational video that patients with prostate cancer could watch to receive information about genetic testing. More than 70 percent of patients chose the video, and 94 percent of them proceeded to genetic testing. This was the first published patient-choice study regarding prostate cancer genetic testing.

“Thinking of novel strategies to deliver genetic information and access to genetic testing is very exciting to me,” she said. “I plan to bring innovative models across cancer types so we can have an integrated effort to raise awareness across the diverse and underserved populations served by Yale, and ensure that standard-of-care genetics services are available across all clinical settings in the Smilow Cancer Network. That’s one of my primary goals.”

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