Empowering Patients & Families with Knowledge

Even though Kirsty Harris was young and had no personal history of cancer, she decided to pursue genetic counseling due to a strong family history. Her mother was diagnosed with breast cancer before the age of 50, and Kirsty herself had been found to have atypical breast cells. In 2015, when Kirsty and her husband moved from Australia to the United States, she knew the importance of finding care and continuing to be followed. After speaking with her doctor, she was referred to the Smilow Cancer Genetics & Prevention Program at Smilow Cancer Hospital, where she met with genetic counselor Jessica DiGiovanna to discuss her risks and what genetic testing would mean for her. The Smilow Cancer Genetics & Prevention Program is an interdisciplinary team that includes geneticists, genetic counselors, physicians and nurses who work together with the goal of providing cancer risk assessment and taking steps to prevent the development of cancer.

After watching her own mother endure extensive treatment for stage three breast cancer, Kirsty jumped at the opportunity to have a say in her own health and future at the age of 30. In addition to testing for the more commonly known BRCA gene, it was decided that Kirsty be tested for an extended panel of genes, including a lesser known gene, CDH1, which is associated with a higher risk of both breast and gastric cancers. To everyone’s surprise, Kirsty tested positive for CDH1 and negative for BRCA. The discovery of this rare gene indicated that Kirsty had a significantly increased risk of developing stomach cancer, even though no one in her family had any form of gastric cancer.

“I had prepared myself for the possibility that I would test positive for BRCA and need a double mastectomy, but nothing could have prepared me for the news that I was also at risk for stomach cancer,” said Kirsty. “Thankfully the team at Smilow was prepared for action, and I met with Dr. Xavier Llor within a week of receiving the results.”

Xavier Llor, MD, PhD, Associate Professor of Medicine (Digestive Diseases) and Co-Director of the Smilow Cancer Genetics & Prevention Program, commented that it was an unusual finding due to the lack of family history. After going over the results and explaining the high risk for stomach and breast cancer with Kirsty, it was decided that an upper endoscopy would be performed with multiple biopsies taken before a removal of the stomach was considered. Out of the 100 biopsies taken from the stomach, Kirsty was found to have a small foci of cancer cells in one of the samples, or stage I stomach cancer. The cells found were consistent with the theory that the CDH1 mutation was the cause.

“The difficult decision of whether or not to have my stomach removed was therefore made for me,” commented Kirsty. “I would have probably delayed the surgery until my children were older, which would have made for a very different story. This was like finding a needle in a haystack. Finding these tiny spots of cancer in my stomach has led to...”

Kirsty and her daughters, Ruby and Georgia
Dr. Llor commented that most often stomach cancer is diagnosed in more advanced stages and that it takes a combination of a high level of knowledge of this rare condition and the proper expertise in genetic testing to create these success stories. “A team approach has been critical to Kirsty’s care,” said Dr. Llor. “With the recent generalization in the use of genetic panels, more and more cases are being diagnosed that would not have been suspected using the standard clinical criteria, and therefore, would not have been tested for.”

Charles Cha, MD, FACS, Associate Professor of Surgery (Oncology and Gastrointestinal), performed the total gastrectomy with little complication. He was able to perform the procedure laparoscopically, which decreased Kirsty’s recovery time. Dr. Cha is one of only a few surgeons in the region that has considerable experience in laparoscopic and robotic surgery for gastric cancer. Kirsty commented, “I was extremely lucky to end up in this part of the world with such a highly skilled team right at my doorstep. It made it so that I could get back to caring for my family; I was sitting and playing with my baby girls the very next day and I only have a small scar below my sternum.”

With her recovery well underway, Kirsty met with Erin Hofstatter, MD, Associate Professor (Medical Oncology) and Co-Director of the Cancer Genetics and Prevention Program to focus on her increased risk of breast cancer. Almost a year after the surgery to remove her stomach, she underwent a double mastectomy and was thankfully found to have no sign of cancer.

“My only question for Dr. Hofstatter was how soon we could start the surgery. I chose this path to give myself the best chance at life and I wanted to do it in the most effective and safe manner possible. My daughters, Georgia and Ruby, are three and two years old. So far we have managed to stay one step ahead, thanks to the team at Smilow, and I plan to be here for my girls for a long time.”

Dr. Nina Horowitz performed the mastectomy and Dr. Henry Hsia immediately scheduled her for reconstruction in an effort to provide seamless care. Kirsty’s mother, who is still living in Australia, decided to undergo genetic testing as well, and was found to carry the CDH1 gene. Looking back at the family tree on her maternal grandfather’s side, now knowing what to look for, there were several unusual deaths that could have been related to the gene, although nothing can be confirmed. Her mother had her stomach removed, which ended up revealing a small amount of gastric cancer. She also chose to have her other breast removed, which revealed early stage lobular cancer. Her uncle, who also tested positive for the gene, is scheduled to have his stomach removed later this year.

“The next challenge, far greater than what I have faced so far, is that genetic testing now awaits my daughters when they are older,” said Kirsty. “There is a 50% chance that they will carry the gene, but in that they have their mother and grandmother to guide them and to be an example of how strong a person can be and that with medical intervention, we can choose our own future. I truly hope they have a mother I did on their side, fighting for them every step of the way.”

Living without a stomach has been difficult for Kirsty, but far easier than the alternative. She commented that she never felt pressured to make a decision, but rather empowered at every stage, starting with the initial decision to undergo testing. And while Kirsty does worry about her future and the challenges to come, having a future to worry about is worth the struggle.