Prevention Matters

| FOX CHASE CANCER CENTER RISK ASSESSMENT PROGRAM PUBLICATION | FALL/WINTER 2024 |

"A bad day post-op is still better than a good day with cancer"

Tina Bucci, a carrier of BRCA2 and MRE11A mutations

I am a 39-year-old stay-at-home mom to four kids. In June of last year, we found out that my mother had pancreatic cancer. I became her primary caretaker and in my research into treatment options, I discovered a possible genetic link between pancreatic and breast cancer. My grandmother passed away in her 30s from breast cancer. One cancer is bad luck, but two in the family tree was enough for me to pursue genetic testing.

My mother and grandmother passed away too young from cancer without the benefit of genetic testing. I want to be around for my kids and the best way to show up for them is to take responsibility for my health. I felt like I owed it to my children to find out our family risk. This is something that each of them will have to face in time and I hoped to set a strong example of survival.

My two sisters and I all tested positive for BRCA2 in January. An MRE11A mutation was also identified at the same time.

What was your reaction to these mutations?

I fully expected to test positive, but I was still shocked. My results were available through the online portal of the company I tested with. I had the opportunity to gather my thoughts, write down questions, and do research prior to my appointment. I felt like I was better able to absorb the information at my appointment because I had time to prepare.

What preventative measures have you undertaken due to your BRCA2 mutation?

Since testing positive I have received a prophylactic bilateral mastectomy with reconstruction. While I was counseled on the option to have high-risk screening instead of the mastectomy, I wanted the security of the surgery. Before the procedure, my odds of developing breast cancer were greater than winning a coin toss. Now, my odds are even less than that of the general population. I might have hesitated if I were younger but having breastfed each of my children for 2 years, I felt like they had "served their purpose". I wouldn't hesitate to do it all over again.

Have you found any support groups that were helpful?

My sisters and I all tested positive together, we've formed our own mini-support group. Before my surgery I did find some great resources online. **BRCASTRONG** sent a completely free care package that included a shirt with drain holders, my favorite post-mastectomy bra, and even a sweet stuffed animal for my kids to cuddle when they couldn't climb on me.

Pockets of Hope was another organization that also sent me a free hoodie with drain pockets. It was nice to have that support because unexpected surgery can get expensive, not just in copays, but in all the recovery things that insurance doesn't cover: wedge pillows, compression garments, button-down pjs. It seems silly, but it can add up.

If I had anything else to add it would be this:

- A bad day post-op is still better than a good day with cancer.
- Instead of being devastated by my misfortune in the genetic lottery, I consider myself lucky to have the opportunity to act and be proactive.
- People worry about telling their kids more than they should. I have children ages 4-13 and was honest with all of them from the get-go. When you're honest with them it becomes a normal part of life.

"Have faith and lean on your people"

Natalie Clee, a paraganglioma patient and a carrier of a SDHB mutation

For nearly five years before my diagnosis, I struggled with persistent back pain. Being young and healthy, my doctors initially prescribed medications to alleviate my symptoms. I sought relief from massage therapists, chiropractors, and physical therapy, but the pain never subsided. Eventually, I was referred to an orthopedic doctor who ordered some imaging.

On March 17, 2023, at the age of 45, I was diagnosed with cancer. I was referred to Fox Chase Cancer Center and after a series of tests, scans, and a biopsy, I was diagnosed with a rare cancer called Paraganglioma. They found tumors on my spine, skull, pelvis, hip, and other areas.

The largest tumor, which had caused pain for many years, was removed in August 2023. The remaining tumors are closely monitored with scans every three months.

During that same year, I underwent genetic testing at Fox Chase, which revealed an SDHB gene mutation. This mutation is known to increase the risk of developing certain cancers, including Paraganglioma.

What was your first reaction to cancer diagnosis?

I was mostly shocked. Cancer hadn't affected anyone in my immediate family. The genetic testing process opened the door to conversations within my immediate and extended family and we started learning more about our genetic makeup.

A few years prior to this, my younger brother had a gastrointestinal stromal tumor (GIST) removed, which doctors labeled benign. He didn't need any follow-up treatment, so we didn't think much more of it at the time. However, he had been tested and was SDHB positive. I believe knowing this at the time of my diagnosis helped to pinpoint my Paraganglioma and speed up the treatment process for me.

What was your reaction to your positive genetic testing results?

My results were not surprising, since at that time we had already started reviewing my brother's test and learning more about the mutation. What was surprising and almost unbelievable was when a second cousin of ours had a tumor removed from his adrenal gland (Pheochromocytoma). It prompted many extended family members to also be tested. Many of them have discovered they also carry the SDHB gene mutation and have subsequently been scanned. The awareness has been invaluable.

Who is your support system?

My greatest support comes from my husband, who has been a champion for my health from the start. He has attended almost every appointment and has always listened and advocated for my care. My mother-in-law has been a true blessing and has been with me during some of my toughest days. She is a retired nurse, so her background in the medical field really helped when I needed an extra ear listening to the doctors. My family and friends were also a huge support emotionally and even financially as we figured everything out. I couldn't have done it without them, and I am beyond grateful for their support.

How did you maintain emotional and mental balance during these difficult times?

Trying to stay positive and grateful helped to balance things for me. I found if I thought of something I could be thankful for, it helped with depression. It's hard to be depressed in a state of gratitude. I started a prayer journal and found it helped me calm some of my anxieties and fears. Music and sleep meditation helped when I needed rest. Giving myself grace and learning my new limits and accepting a new "normal" is a work in progress, but I keep moving forward.

What would you like to say to other people fighting cancer?

Have faith. On days when I felt weak, my faith gave me strength. On days when the uncertainty overwhelmed me, faith brought me hope. It's easy to let fear take over during times like these, but faith brings peace. Lean on your people. I learned to ask for help when I needed it and sought resources that could make things a little easier.

"One mutation, two mutations, three mutations"

Bonnie Jacobs, a carrier of BRCA2 and PALB2 mutations

I grew up in Elkins Park, PA which is about five minutes from the Fox Chase campus. My mom was diagnosed with melanoma when I was 16 and then with breast cancer a few years later. Because I was so young when my mom was first diagnosed, Fox Chase was very unthreatening to me. I thought of it as the neighborhood hospital that helped people.

My mom was a very proactive person. She has a strong family history of cancer, including a first cousin who was diagnosed with ovarian cancer at a young age. She found out about the Family Risk Assessment Program at Fox Chase and signed my family up for it. My mom, sister, and I (along with our first cousins) underwent genetic testing in 1998, but my sister and I did not opt to get the results immediately. I didn't worry that much about cancer because all of the cancer cases were on my mom's side, and I resembled my father's side of the family. It was only after my sister was diagnosed in 2002 with early-stage breast cancer (and confirmed to have the BRCA1 mutation) that I got my test results. I was correct about taking after my father, but I hadn't considered that he would also have a genetic mutation. While my mom and sister had BRCA1, my father and I were carriers of the BRCA2 mutation.

My sister and I both underwent mastectomies and reconstruction and prophylactic oophorectomies. I completed my surgeries in 2009 and, for the next 11 years, did not do much in terms of surveillance aside from my yearly gynecologist appointment. When my sister had a reoccurrence of breast cancer in 2020, her doctor suggested that we undergo updated genetic testing. We were both found to have a mutation in the PALB2 gene, which increases risk for breast, ovarian and pancreatic cancer.

What was your reaction when you recently found out about the second mutation?

We're a pretty pragmatic family. We know that cancer is part of our life and we're grateful that we have access to resources that have allowed us to have the best preventive care and treatment. I never had a moment when I mourned the loss of my original breasts or ovaries or a "why me" moment because I carried genetic mutations. I was much more focused on the fact that I had options and on doing what I could to reduce my risk. I will admit that the increased risk of pancreatic cancer does scare me...but, again, I try to be grateful. The Risk Assessment Program continues to be there for me and connected me with Dr. Malorie Simons who I see for annual pancreatic screening.

Why is it important to you to share your story?

You can only help people if you talk about it. I have always been open about my own journey and my family's story. My sister and I feel lucky that my mom was so proactive to get us involved in the program when it was still in the early stages. It's more common now for people to talk about cancer and genetics, but it wasn't the case 27 years ago.

I recently read a story about a woman who has familial ALS and chose not to have genetic testing because there is not much that she can do to prevent developing ALS. Her decision is understandable. But for my family, we could act. Thanks to Fox Chase, instead of having something scary hanging over our heads, we have had access to the best medical team and scientific resources who have been helping us "do something about it" for almost three decades.

"My BRCA 2 mutation is the biggest gift that saved my life"

Allison Perlis, a pancreatic cancer survivor and a carrier of a BRCA2 mutation

I was 36 years old when I was diagnosed with pancreatic cancer. I went to the doctor because my eyes looked yellow, and I was itchy all over my body. I went to a dermatologist first, and then to my primary care doctor to get some blood work done. After seeing elevated bilirubin levels, a CT scan was done and found a mass on my pancreas.

Through my primary physician I underwent BRCA1/2 testing for 3 common genes in Ashkenazi Jewish population. My results came back negative. I started receiving a standard chemotherapy but that didn't work for me; the cancer spread to my liver during the first months of my treatment. The cancer progressed to stage IV and I was no longer eligible for surgery.

I met Dr. Michael Hall through a family member who worked at Fox Chase. He suggested that I undergo additional genetic testing. Through this testing I found out I have a BRCA2 mutation. This opened a door for a new platinum-based chemotherapy targeting tumors with BRCA2 mutations. At this time, I was collecting a lot of information and researching about clinical trials. I heard about the PARP inhibitors for BRCA2 related cancers being used for breast and ovarian cancer, but not pancreas at the time. We brought it up initially, but it wasn't something to consider, it wasn't a 1st line treatment yet. After several months on FOLFIRINOX with no new growth, it was time for a break and I asked to try taking a PARP inhibitor, a medication called Lynparza. The treatment worked and this made me a candidate for surgery. I have been cancer-free for 6 years now.

What was your first reaction to cancer diagnosis and the BRCA2 mutation?

My cancer diagnosis was a shock to me. I am an eternal optimist and felt that I was going to get past this, and it was just another bump in the road.

I was surprised to find the BRCA2 mutation, because there is no history of any genetic related cancers in my family. I did discover my father was adopted, and my mom's parents died young, leaving me with very little information regarding my parents.

Having a mutation was the biggest gift that saved my life. I choose not to be afraid of the mutation. I think that knowledge is power in my case. It allows me to be proactive in protecting myself. I don't view having this mutation or a cancer diagnosis as a death sentence. I think that you must look at life as a journey.

How did you explain your cancer diagnosis to your young children?

I didn't want it to be a shameful secret, I was honest and direct with them. They were so young and didn't fully understand the risks. As they have gotten older, they're amazed that all of this happened when they were so little and that I'm still here. It's a big blessing.

How did you maintain emotional and mental balance during that difficult time?

It took a great amount of support from my family, stepping in and helping me with the kids. I also prioritized yoga and meditation and tried living in the moment. This really benefited me in keeping my mind and body balanced while going through cancer treatment. I think that helped me not be nervous about the future and allowed me to still be present in my life during that time.

Speaking in support groups about my feelings helped me a lot. And now, as a survivor, I'm able to talk to people who are newly diagnosed, which is very meaningful to me. Genetics played such a huge role in my treatment. It is important to me to share that knowledge.

What would you like to say to other people fighting cancer?

I always listen to the doctors and would think that that was the only solution. But sometimes you need to think outside the box, advocate for yourself, and take some risks. It's okay to ask for what you want. Not everyone feels that way when they're on treatment. I would encourage people to ask for anything that they think could be helpful and do their own research.

Find ways to be hopeful. Look for things that you can feel positive about and try to focus on those. There are a lot of beautiful stories out there and there's a lot of hope.

"Don't ever give up. There is light at the end of the tunnel."

Janice Hudnell-Davis, a pancreatic cancer patient and carrier of an ATM mutation

I was diagnosed with pancreatic cancer in January 2022 at 44 years old. I went to my family doctor because I thought that I was suffering from hemorrhoids. My primary care doctor ordered tests including a CAT scan that showed a spot on my pancreas. I was immediately referred to Fox Chase Cancer Center. I was initially very nervous, but once I walked through the doors and met my awesome medical team, Dr. Reddy, Dr. Hall, Dr. Meyer, and Eric Tetzlaff, I felt relieved and comfortable.

After eight cycles of chemotherapy and 25 rounds of radiation my scan showed that the mass had shrunken. I underwent my Whipple surgery in December of 2022. I will be cancer free for two years in December, and it feels amazing!

What was your feeling when you heard about your pancreatic cancer diagnosis?

I was in disbelief. I lost my birth father in 2020 to gallbladder cancer and my bonus father to pancreatic cancer six months prior to my diagnosis. My mom was devastated. I couldn't understand how and why it was happening to me. I was blessed with my medical team, and my support system: my husband, my children, my mother and my siblings, and a lot of close family and friends.

How did you find out about your ATM mutation?

Dr. Hall ordered a genomic test on my tumor. The report identified a mutation in the ATM gene. Then I met with a genetic counselor who explained the risk and screening recommendations for me and my family. When the results came back positive, I kind of knew that it came from my father. My mom underwent testing to confirm this. I shared my genetic information with my siblings so they all can be aware.

How has having an ATM mutation changed your screening and medical care?

In addition to my pancreatic cancer follow up visits, I meet every 3 months with a genetic team at high-risk breast and ovarian clinic. The ATM mutation puts me at higher risk for both breast and ovarian cancers. I want to be aware of everything that can help me prevent other cancers. For my breast cancer risk, I get my MRI with the regular breast exam and mammography. I also do more screening for ovarian cancer risk.

So why is it important to you to participate in research?
I gladly agree to participate in research offered to me at Fox Chase. Research is extremely important; doctors and researchers learn new information to give hope to other people that are going through the same situation. I also do it for myself, so I can have a better understanding of my condition.

I wanted to say to other people diagnosed with cancer: "Don't ever give up. There is light at the end of the tunnel."

MARK YOUR CALENDAR

Together Facing Pancreatic Cancer 2024

Date & Time: Thursday, November 21, 2024, 05:30pm to 08:00pm
Location: Fox Chase Cancer Center Cafeteria Center Building
333 Cottman Avenue, Philadelphia, PA 19111

Living with Lynch Syndrome: A Genetic Counselor's Point of View

Allaina Brock, a carrier of an MSH2 mutation

My name is Allaina Brock, and I am a certified genetic counselor who graduated from UPenn's Genetic Counseling program in May 2024. I had many insightful rotations in graduate school including in prenatal and pediatric specialties, but I was especially enthusiastic about my two cancer rotations.

I was diagnosed with Lynch syndrome 3 years ago, at age 22. I don't have a personal history of cancer but decided to undergo genetic testing due to a family history of cancer and known MSH2 mutation in the family.

Do you recall how you felt when your family shared the risk of Lynch syndrome with you?

I don't recall the exact moment I began to fully understand the impact of Lynch syndrome or how I felt about it. However, one moment in particular that I vividly recall, is when the gravity of Lynch syndrome seemed to hit my younger sister. After learning all the risks associated with Lynch syndrome in my cancer genetics course, I was having a discussion with all of my siblings about what I had learned. After going through all the risks, one of my younger sisters had become extremely quiet and teary-eyed. I felt terrible.

In a counseling setting, I would be careful to ensure that patients are processing information effectively, but with my family, I was so eager to share what I had learned that I overlooked the need to gauge their emotional response. Learning about an elevated risk for cancer can feel like a bomb has been dropped on you. Processing new information can be overwhelming and difficult to absorb or hear anything else.

From a young age, I was aware of family members' Lynch syndrome status and cancer journeys. The discussion about Lynch syndrome was always handled openly in my family; however, I cannot pinpoint a time that I was sat down and educated about my personal risk.
Reflecting on my own experience, I believe a more structured conversation with my parents about Lynch syndrome would have been beneficial for me and my siblings.

To this day I am the only one of my siblings who has been tested for Lynch even though I am certainly not the oldest. I suspect that their understanding regarding the seriousness of Lynch syndrome might be limited due to the casual way it has been discussed in our family.

How has having Lynch syndrome changed your medical care?

Having Lynch syndrome has emphasized the importance of being a self-advocate for your own medical care. I am fully aware of the cancer screening recommendations for myself given my Lynch syndrome status and current age. However, just because I tested positive for Lynch syndrome does not mean I have any support reminding me and helping me navigate how to get the proper screening. It is so important to find a care team that you can trust and that will help guide you through your preventative screening and management.

What made you decide to participate in the Lynch syndrome vaccine study at Fox Chase?

Knowledge is power. Knowing I have Lynch syndrome gives me a certain level of power over my health. With this awareness, I can take proactive steps to ensure I receive proper screening and potentially catch cancer early if it develops. Following screening guidelines is crucial, but screening alone is not a treatment.
The introduction of a potential vaccine represents a groundbreaking advancement in cancer prevention. I am enthusiastic about the possibilities this could offer for managing Lynch syndrome cancer risks in the future. I was eager to participate in this clinical trial to contribute to this promising development and would be thrilled if it proves to be effective in preventing cancers and becomes accessible to all individuals with Lynch syndrome.

"I'm here today because of Fox Chase's commitment to research."

Jonathan Miller, a prostate cancer patient and a carrier of an ATM mutation

I was diagnosed with prostate cancer in 2010. The cancer spread to my hip, spine, sternum and skull. In all of the ups and downs of this illness, I appreciated the ability to also maintain a sense of humor in it all. I was given an extra scan of my skull and it was apparent that I had a very, very thick skull (my wife could have told them that), and therefore not to worry about that part of the cancer!

The medical team was supportive and direct when I asked how much time they thought I had. I was told they were going to do all they could to get me to 2015. Here I am 14 years later, I can only be thankful. A big part of that thankfulness is because of Fox Chase's commitment to research. I was included in a clinical trial utilizing four radioactive infusions, which attacked cancer cells and for the past two years kept me free of cancer growth. Every two months I come to Fox Chase for further tests to monitor the ongoing effectiveness of the treatment.

Recently, I was offered the opportunity of pursuing genetic testing which could be used to potentially help others in my family. I learned that I do have an ATM mutation. It has become clear that my daughters and their children may be affected by this mutation too. I wasn't sure how to tell them. Part of me says "of course they should know, so they can take preventive action", and another part of me wants to be very careful not to provide information that may not be welcome at this time. I am very grateful for my genetic counselor who gave me much room to process at the speed I was comfortable with.
I told my daughters and my siblings about the genetic results and now it's up to them what to do with this information.

I have been called a 'responder' to the treatments I've received. I attribute my success as a responder to my medical team, Dr. Geynisman and Victoria Wong, the research and the benefits of treatment, an increased commitment to be physically active giving my body a chance to battle this illness, and a grandson who never finishes a prayer without saying, "and dear God, please take away Pop's cancer."

May God bless Fox Chase and all they serve so well - even more!

INDEX mutations from the patients' interviews

ATM - Increased risk of breast, prostate and pancreatic cancer, and elevated risk of ovarian cancer.

BRCA1/2 - Increased risk of breast, ovarian, prostate, melanoma and pancreatic cancer.

Lynch syndrome - Most common hereditary cancer syndrome is caused by a mutation in MLH1, MSH2, MSH6, PMS2, and EPCAM genes. People with LS have a high risk of uterine and colorectal cancer, and elevated risk of ovarian, urinary tract, stomach, small intestine, pancreatic, biliary tract, and skin cancer. Certain cancers are developed before age 50.

MRE11A - A person with one MRE11A mutation does not have an elevated risk of cancer. However, a person who inherits two MRE11A mutations may have a pediatric onset syndrome called Ataxia telangiectasia-like disorder (ATLD), a rare neurodegenerative disorder.

PALB2 - Increased risk of breast and pancreatic cancer, and elevated risk of ovarian cancer.

SDHB - Increased risk of Paragangliomas (rare neuroendocrine tumors affecting the head, neck, upper body or abdomen), renal cancer, and gastrointestinal stromal tumors (GIST).

- Both men and women can inherit and pass on a gene mutation
- People with mutations may develop one or more cancers, or they may not develop cancer at all.

TOGETHER FACING

Hereditary Cancer

TUESDAY, NOVEMBER 12, 2024 | 5:30 – 8:00 PM

VISIT **DONATE.FOXCHASE.ORG/TOGETHERFACINGHEREDITARYCANCER** TO REGISTER

Fox Chase Cancer Center
Temple Health

All patients, survivors and caregivers are welcome to attend this educational and inspirational program, which will cover genetic risk for Breast, Colon, Pancreas, Prostate, Kidney, Uterine, Endometrial and Ovarian cancers.

SCAN ME

Together Facing Hereditary Cancer
Tuesday, November 12, 5:30 - 8:00 PM
Fox Chase Cancer Center Cafeteria and Auditorium
Register: https://www.foxchase.org/events/together-facing-hereditary-cancers-2024

Editors: **Yana Chertock and Deborah Grace**
Design: **Joe Gillespie, 4545 Creative**

Risk Assessment Program
333 Cottman Avenue
Philadelphia, PA 19111-2497

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