

## A Family's Story: Allen, Judy, Sarah and Lea

**A**LLEN is 52 years old, married, and the father of two daughters. His mother died of breast cancer when she was 49 years old. Allen was in Grade 7. His sister died of ovarian cancer at the age of 54. A cousin was diagnosed with breast cancer at 37 and ovarian cancer at 55. There were other cancers too, among them pancreatic, colon and prostate. A BRCA2 gene mutation was found in Allen's family so he decided to have genetic testing to find out if he was a carrier.

*In this issue of Gene Scene, Allen\*, his wife Judy\*, and their two daughters Sara\* and Lea\* talk about their experiences with genetic testing.*

*\* Names have been changed.*

**CANCER IN THE FAMILY** Cancer was mostly associated with the females in our family. I didn't feel particularly susceptible, but it was always in the back of my mind. When we found out that it was hereditary, it wasn't a big surprise. It was an explanation of what intuitively we suspected.

**MY TEST** I wanted to be tested to hopefully rule out the likelihood for my daughters. I had a 50/50 chance of testing negative. But if I tested positive, my daughters could make whatever choices or decisions that might be beneficial to them.

I took the test and waited a few months for my results. I didn't fuss much about it. I suspected the worst before I went in but when I found out that I was, in fact, a BRCA2 carrier, I was disappointed. I had hoped for a different outcome. It would have been better for my daughters.

**DISAPPOINTMENT** When Sara tested positive I was disappointed again. I thought, sooner or later somebody has to get a break. Obviously each generation hopes to get off the hook. Mom must have been a carrier, most of my siblings are carriers, and now, one of my daughters is.....It seems the 50/50 odds aren't quite as fiftyish as we thought.

**RISK** When you're a parent and you've got teenage children you're glad every time they get home safe. Genetic risk for cancer is only one risk dimension and it's a "down the road" risk. It is one piece that you can do something about to try and lower the risk. There are so many other factors that nobody can predict or have influence over. To me this is one piece. It's a thing you learn about, take whatever precautions you can that make sense, and then get on with living.

**HOPE** I'm still hopeful that advances in medicine and science will make more options available. The years do slip by pretty quickly though. When we first became aware of this, I thought there was a ten, fifteen or twenty year window for our kids. And now one of them is 25 and one is 28 and the window is shrinking. Time is moving forward fast and I don't know if the progress in the cancer research is happening fast enough.

**PASSING ON THE GENE** How do I feel about passing this gene on to my daughter? It's a card that was dealt to me. I think in years to come almost everybody's going to find that they've dealt certain genetic codes to their kids. Some are good some are bad. This is one that's been identified but there will be more. If there was guilt there's also pride in passing on some pretty good traits to the kids.

**S**ARA is 28 years old and the eldest of Allen and Judy's two daughters. She and Michael got married two years ago. Recently she learned that she inherited the BRCA2 gene mutation from her father. Her sister Lea is still waiting for her genetic test result.

**DAD'S RESULTS** Dad told us about his results as soon as he got them. I was 24 at the time. I was not as concerned as my sister was. It was something that seemed very much in the future for me.

**TELLING MY BOYFRIEND** I told Michael well before we were even engaged. I just told him that there's a crazy family history of breast cancer in my family. My dad carries the gene that indicates whether or not I would have a high risk of developing breast or ovarian cancer and I have not yet

been tested for that. It is a very good possibility that sometime later in my life I may develop breast cancer or would chose to prevent that by having a mastectomy. He was fine with that. It didn't feel as real then but he's been supportive throughout it.

**DECIDING TO TEST** Lea and I talked it over and we both agreed that we'd rather know than not know. I felt that was better than getting hit when I was not expecting it. You can compare it to a tornado. If they could predict tornados ten years in advance, you have time to prepare, make arrangements, or move before it hits. On the other hand, if you've got 45 minutes and the storm warning on the radio you have a lot less time to make a careful decision. It's like that with the possibility of having cancer. I've got time to prepare. It's not a death sentence.

**GETTING MY RESULTS** The doctor at the Genetics Clinic did a quick overview and then she said, "You do carry the gene." I wasn't emotional. It really didn't bother me. To some extent I don't understand why it bothers other people. It's something you have carried for your whole life. Nothing has changed in your body. You're just more aware of it.

**PLANNING AHEAD** It was a fleeting thought that I may chose not to have children but when I really thought about it that's an awfully tough choice to have to make on behalf of Michael and my parents. There is only a 50% chance of passing the gene on to them. Our children should be able to make their own choices, as we have. We plan to have kids over the next couple of years.

Once our family is complete, I will probably choose to have preventative surgery. I'm leaning that way right now. I'm not scared of getting cancer; I'm scared of the process we have to go through to not get cancer. In the meantime, of course, I will do all the necessary screening. I don't think there'll be a magic pill that I can take in ten years. I wish I could think that but I don't.

**MY SISTER** I truly hope she's negative. If it had to be one of us, I'm glad it's me and not her. If she does carry it, I'll be sad for her but at the same time we'll be in this together.

**L**EA is 25. She knows that her sister tested positive and now she's waiting for her own test result.

**DAD'S RESULTS** When my parents first told me dad was positive I didn't get upset. It didn't really affect me right then; I was only 21 years old. I kind of forgot about it. But then it would come back and I'd think, "I could have the gene. Fifty/fifty is a pretty serious odds for me." Sometimes I thought it wasn't very fair that I had to deal with it at that age.

**DECIDING TO TEST** As I got older the desire to know grew. When I saw the genetic counsellor, she talked about a "sense of control" I think that was the perfect way to describe it. I have more of a sense of control if I know one way or the other.

Sara went into testing in a different mental state than I did. She had the "I am positive" attitude. And when she got her results, she was very frank about it. I don't expect to be positive. Before I got tested, I'd sometimes play out the two scenarios...I might be....I might not.....I'd flip back and forth. That's what I didn't like; I didn't like hoping that I'm not positive. There still may be a hammer that comes down and I'd rather have it now.

It wasn't a hard decision to get tested. At first I was concerned about insurance. But it was really easy for me to get insurance at my age. It wasn't expensive and they didn't ask medical questions.

After having the test I was relieved. Now I don't think about whether I'm positive or negative. I'm just wondering when the clinic is going to call.

**SISTERS** When I heard that Sara got tested, I thought that we both would have to go to the Genetics Clinic together. We are two very different people but there is something unique because we are sisters. With sisters it's more real. The hardest thing for me was when Sara got her results.

**WHEN I GET MY RESULTS** I think I'll go by myself. My mom would get upset. And my dad...I think it's hard to see your daughters go through this when it's your genes. I think he feels the pressure of it even though he couldn't do a thing about it. I don't want him to have to be there and see me upset, if I get upset.

If I'm negative I'll be relieved, happy. But also a little bit guilty I think because my sister is positive. I know she'll be happy for me but I can imagine, in ten or twenty years, if she's going through a tough time and I see her going through it, I think I'd feel guilty.

If I'm positive I can make a plan of action and take more of an active interest in the research. I would look at my options and what I could be doing now to protect myself.

Eventually if I get married maybe I'll want to have kids right away. It just depends how my life goes what options I chose. But on the other hand, knowing prepares me for the decisions I'll have to make far down the road.

***JUDY** is Allen's wife and the mother of Sara and Lea. She learned about the family history of cancer when she met Allen 30 years ago, but never thought her daughters could be at such high risk.*

**BIG NUMBERS** When I first heard the news of Allen's positive result, my first reaction was emotional. I was concerned for our daughters. And sad. The numbers are so high, especially for the girls. They say there is a 50 – 85% lifetime risk of breast cancer for women who are carriers. Those are big numbers in the percentage game. Later, I concentrated more on what they could do to be proactive. I wanted to know first what problems they might face. I wanted information.

**SINCE SARA TESTED POSITIVE** I've been worried. I think about the fact that she hasn't had her children yet and about all the decisions she's going to have to make along the line. I have a little bit of hope that there will be some sort of medical intervention possible instead of surgery. I would support her in whatever she does. And I'm anxious to hear what Lea's results are.

**ADVICE TO OTHER FAMILIES** I would say be as knowledgeable as you can. It's also important to communicate. One person in our family started the ball rolling and communicated this to everyone. That helped us to understand what was going on and what should be done. I think also of value to families is having a doctor who's interested in keeping up on the latest information.

**GENES** I can't blame Allen for bringing this into the family because I'm in the same situation. My heredity is early heart attacks and thyroid disease. I happened to get the thyroid disease that my dad had. It's the way life is. We can't feel guilty over it. The genes are the way genes are.

**SUPPORT GROUP** It's good to know there is a support group. It is just nice have a number to phone and be able to get information and to hear about other people's experiences.

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***Post Script:** Since then, Lea got her results and she tested negative. She was relieved to hear that she does not carry the BRCA2 gene mutation that runs in her family. Now she knows she is not at high risk and she will continue to follow the normal surveillance recommended for women in the general population.*