



CHAPTER 9 : PATIENT STORIES & CONSUMER PROFILES

This chapter contains four stories about inherited cancer, newborn screening, late-onset disease and family history told from the perspective of a patient or consumer. For some patients, diagnosing a genetic condition can be a challenging and lengthy process involving many doctors and office visits, examinations, testing, and months or years of stress and uncertainty. For other patients, the lack of treatment or effective interventions can prove to be extremely frustrating and difficult to comprehend. These stories can help both health professionals and patients understand the issues faced by patients and families affected with a genetic condition and how they overcame and dealt with these issues.

9.1 INHERITED BREAST AND OVARIAN CANCER

At a holiday dinner, my family and I were talking about my sister's recent diagnosis. Just 38 years old, Rachael had detected a lump in her left breast and was diagnosed with breast cancer. She was recovering from the mastectomy she had chosen to have in order to increase her chances of survival. It was still too painful to talk about our mother's death, years ago, of breast cancer at the age of 48, but from the silence in the room, it was obviously on everyone's mind. It was also believed that a great-aunt had died of some form of cancer, although no one was really certain.

The high incidence of cancer deaths in our family was cause for concern. My husband and I were considering having another child. We had two healthy sons but wished for a little girl. I was only 34 years old, but what if I developed breast cancer in 10 years? Would I be able to care for my children? Could I pass on the risk of breast cancer to my children?

I met with my family doctor to discuss my family history of breast cancer. I asked if there was any way to find out if I was at risk for the disease and if there was anything that could be done to prevent the disease from occurring. My doctor referred me to a genetic counselor who specialized in inherited cancer to discuss my risk for cancer and the types of genetic testing available. The genetic counselor told me that a small percentage of all breast cancers are inherited and that two genes, called BRCA1 and BRCA2, have been discovered to cause inherited breast cancer in some families. The genetic counselor also told me that genetic testing is available to some women with a family history of breast and/or ovarian cancer. A positive result would indicate that I have a much higher risk of cancer, but not a 100 percent certainty.

My husband and I were overwhelmed by this information and felt that we needed to talk to someone who had experienced this situation. We found a support group in our community and spoke with the director, who also had a family history of early-onset breast cancer and had decided to undergo genetic testing herself. After several weeks, and with my family's support, I decided to undergo genetic testing. My sister Rachael was tested first for specific mutations in BRCA1 and BRCA2, and once we knew that she has a particular mutation in BRCA1, my blood was tested. I found out that I have the same mutation in the BRCA1 gene.

9.2 THE VALUE OF NEWBORN SCREENING

We brought our 7lb. 5oz. baby boy home on April 14th. After a tiring but blissfully happy first week of 4 a.m. feedings and little sleep, our pediatrician called to say that one of the newborn screening tests done on the blood spot collected from our son at birth had come back positive. My husband and I both thought that it had to be a mistake; our son Miguel was a completely healthy and happy baby boy.

The positive result was for a disease called homocystinuria. The following week, we took Miguel back to the hospital to have him re-tested. The second test also came back positive. There was no doubt that Miguel had this disorder although he still seemed completely healthy. The doctor told us that children with this rare genetic disorder are unable to break down excessive protein and that in order for Miguel to have a normal life, he would have to be put on a special low protein diet. I had so many questions about what would happen to Miguel. How different would he be from other children? Would his development be delayed? Would he be able to walk and talk and go to school with other kids?

After talking with other parents of children with homocystinuria, several pediatricians, a geneticist at a medical center located two hours away, and nutritionists, we gained some confidence that we could take care of Miguel and provide him with a normal childhood. Miguel



has been on a low protein diet for almost 10 years now, and his disease is under control. He is in the 5th grade and is a very active and bright child. He is doing well in school, plays soccer and baseball, and does all of the things any 10-year old would do: birthday parties, Little League, Boy Scouts. Because Miguel's condition was detected at such an early age, we were able to adjust his diet and prevent symptoms from developing.

9.3 HEREDITARY HEMACHROMATOSIS

Growing up, I was busy and energetic like everyone else. I rarely visited the doctor, and there was no hint of any chronic medical problem.

Soon after I turned 40, I started to notice my joints were achy, but I figured I was just getting old. About a year later, I just wasn't feeling as well as I thought I should. I was always tired, and I had occasional abdominal pain. I saw my doctor for a routine physical. After a long series of tests and visits with specialists, a blood test revealed that I had unusually high levels of iron. A liver biopsy confirmed that I have hereditary hemochromatosis.

To understand my own health risks and the chances of my relatives developing this condition, I met with a genetic counselor and had a genetic test performed.

After meeting with the genetic counselor and doing my own research, I am beginning to understand what it means to have hereditary hemochromatosis. I now know that hereditary hemochromatosis is a fairly common adult-onset condition that can be associated with many serious complications, including heart problems, diabetes, liver cirrhosis and arthritis. I consider myself lucky to have been diagnosed at a relatively young age, before any of the major complications developed. I now have periodic phlebotomies (like donating blood) to keep the iron from accumulating in my body and damaging my organs, and this treatment should allow me to live a long, normal life.

9.4 TYPE II DIABETES

I was 42 years old when I was diagnosed with Type II diabetes. I had had a recurrent skin infection for almost a year, but it seemed minor at first and I had no health insurance, so I put off seeing a doctor. Eventually I noticed that I always felt thirsty, although I was drinking plenty of water and other beverages. In spite of my increased drinking habits and normal appetite, I somehow lost 40 pounds. Finally, the discomfort from the skin condition became so severe that I went to the emergency room, where I was diagnosed with Type II diabetes. It appears I had actually had this condition for some time.

Since my diagnosis, I have learned a lot about my family and about Type II diabetes. I now understand that Type II diabetes appears to be caused by a combination of genetic and environmental factors. My increased risk for diabetes should have been noted many years earlier. If my doctor and I had been aware that my grandfather, mother and two cousins have diabetes, we could have recognized that my risk was greater than that of someone without a family history.

In addition, it would have been helpful to know that my love of sweets and fatty foods and my tendency to be overweight further increased my risk. Being aware of my risk factors might have prompted me to monitor my health more carefully. I could have exercised more and modified my diet, which might have prevented or delayed the onset of my condition or perhaps made it less severe, and I might have acted more quickly when I recognized the symptoms of diabetes. Knowing about your family history can help you to recognize your risk for a condition and possibly enable you to take action to avoid or delay its development.

