



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.

I. INHERITED BREAST AND OVARIAN CANCER

At a family dinner, Samantha and her family were talking about the recent diagnosis of Samantha's sister, Louise. Just 38 years old, Louise 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 their mother's death, years ago, of breast cancer at the age of 48, but from the silence in the room, it was obvious that it was 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 Samantha's family was cause for concern. Samantha and her husband were considering having another child. They had two healthy sons but wished for a little girl. She was only 34 years old, but what if Samantha developed breast cancer in 10 years? Would she be able to care for her children? Could she pass on the risk of breast cancer to her daughter?

Samantha met with her family doctor to discuss her family history of breast cancer. She asked if there was any way she could find out if she was at risk for the disease and if there was anything that could be done to prevent the disease from occurring. Her doctor referred her to a genetic counselor who specialized in inherited cancer to discuss what Samantha's risk was given her family history and the types of genetic testing available. The genetic counselor told her that a small percentage of all breast cancers were inherited. Two genes had been discovered that caused a proportion of inherited breast cancer called BRCA1 and BRCA2. Genetic testing was available to women who had a family history of breast and/or ovarian cancer. A positive result would indicate that Samantha had a much higher risk of cancer, but it was not a 100 percent certainty that Samantha would develop breast cancer.

Samantha and her husband were overwhelmed by the information provided by the genetic counselor and felt that they needed to talk to someone who had experienced this situation. They found a support group in their 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, they decided to undergo genetic testing. Her sister Louise was tested first to identify the specific mutations in BRCA1 or BRCA2, and once they knew that she had a particular mutation in BRCA1, they tested Samantha's blood. Samantha found out that she was a carrier of a mutation in the BRCA1 gene.

II. 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 am feedings and little sleep, our pediatrician called to say that one of the tests done with the blood spot collected from our son at birth for newborn screening came back positive. My husband and I both thought it had to be a mistake and that the samples must have been mixed up. Our son Zachary was a completely health and happy baby boy.



The positive result was for a disease called homocystinuria. The following week, we took Zachary back to the hospital to have him re-tested. The second test also came back positive. There was no doubt that Zachary had this disorder although he seemed completely healthy. The doctor told us that children with this rare genetic disorder were unable to break down excessive protein into other amino acids and that in order for him to have a normal life, he would have to be put on a special low protein diet. I had so many questions about would happen to Zach—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 affected with homocystinuria, several pediatricians, a geneticist at an academic medical center located two hours away, and nutritionists, we gained some confidence that we could take care of Zachary and provide him with a normal childhood. Zachary 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 a very active and bright child. He is doing well in school, plays soccer and baseball, and does all the things any other 10-year would do—birthday parties, Little League, Boy Scouts, etc. By detecting Zachary's condition at a very early age before any symptoms developed and adjusting his diet, Zachary is a bright and healthy boy.

III. ALPHA-1 ANTITRYPSIN DEFICIENCY

Growing up, I was busy and energetic like everyone else. Rarely did I have to go to the doctor. The only real problems I remember were the coughing spells I would occasionally suffer, but I blamed them on my father's smoking habit.

During a routine physical, my doctor noticed high levels of alanine aminotransferase (ALT) on my liver function tests and referred me to a liver specialist. He diagnosed me with alpha-1 antitrypsin deficiency. Though I felt fine, the specialist ordered a liver biopsy and referred me to another specialist, a lung doctor. Everything moved very fast and I didn't understand half of the information I was told and I had no idea what to expect. After a second liver biopsy and a session with a genetic counselor, hours educating myself and my family, I am beginning to understand what it means to be diagnosed with alpha-1 antitrypsin deficiency.

To understand my own health risks and to understand my children's risks of developing alpha-1 antitrypsin, I had a genetic test performed. However, to avoid the risk of genetic discrimination, I have kept the initial genetic testing results out of my medical record.

While it was scary to find out about the disease initially, it has enabled me to possibly change the course of the disease process. For example, I have become a sort of health nut as a result of all of this. I exercise daily, watch my diet, avoid smoky environments and pollutants, and watch my alcohol intake. While everyone should do these things, it is imperative that I do them to help prevent the early onset of disease symptoms. Early diagnosis may have saved me from living a more dangerous lifestyle and damaging either my lungs or my liver. This is one disease where lifestyle changes can result in a longer life when diagnosed early.

IV. TYPE II DIABETES

Sarah was 26 years old when she was diagnosed with Type II diabetes. She had had a recurrent skin infection for almost a year. At first it was a nuisance and since she had no health insurance, it was just something she had to put up with. Then, she seemed to develop an unquenchable thirst and was always drinking water. Despite her increased drinking habits, she managed to lose almost 40 pounds. Finally, the pain from the skin infection became so excruciating that she had no choice but to go to the emergency room. There, they officially diagnosed her with Type II diabetes although she had likely been affected with the disease for some time.

Even without the diagnostic blood tests and a clinical examination, Sarah's high risk of diabetes should have been noted many years ago. Of notable significance, her grandfather, mother, and two cousins have diabetes. Although Type II diabetes is likely caused by a combination of genetic and environmental factors, the high prevalence of diabetes in her family strongly suggested that Sarah was at high risk and a healthy diet and regular exercise could have helped prevent the early onset and severe symptoms she now experiences. Second, Sarah was always a large child and constantly ate foods high in fat and sugar.

Now, at the age of 41, the damage from the disease has devastated the life she was trying to make for herself. Over the past 5 years, she had had two toes on her left foot amputated, her eyesight was rapidly deteriorating, at least four teeth had to be pulled, and she was admitted twice with life-threatening flu infections. Although she refuses to believe it, her doctors have repeatedly told her that just by knowing her family history and a commitment to a healthier lifestyle, much of this could have been prevented.

