This chapter contains four true stories about inherited cancer, newborn screening, late-onset disease, and family history told from the perspective of a patient or consumer. Diagnosing a genetic condition can be a challenging and lengthy process involving multiple doctors and office visits, examinations, testing, and months or years of stress and uncertainty. The lack of treatment or effective interventions can be extremely frustrating and difficult to comprehend. However, genetic diagnosis can enhance educated decision-making and alleviate the stress of the unknown. It can also encourage healthy lifestyle choices and inform family planning. These stories can help both health professionals and patients understand the issues faced by patients and families affected by a genetic condition and learn how to deal with these issues.
9.1 INHERITED BREAST AND OVARIAN CANCER

My grandmother, my dad, and I have all had breast cancer, and our stories are inextricably linked.

My grandmother was diagnosed with breast cancer when she was in her late 30s, had a mastectomy, and lived until age 95! What an inspiration she was!

Most people don't know that men can get breast cancer too. Dad discovered his breast cancer in 2001. While showering, he felt a lump in his left breast/chest area. His doctor confirmed it was suspicious. Dad had a mammogram and then a mastectomy to get rid of the cancer. Shortly after, he was diagnosed with prostate cancer and underwent 40 radiation treatments over eight weeks. His cancers of the breast and prostate were genetically linked. Within a few years, he was diagnosed with bladder cancer. Fortunately, this was caught early and removed, and from that point on he has been cancer free. My dad is now 78 years old and going strong.

After my dad's cancers, his oncologist tested him for the BRCA2 gene mutation and discovered he was positive as a carrier. My siblings and I were tested to see if we were carriers too. I was positive for BRCA2, which carries up to an 85 percent lifetime risk of breast cancer for women.

The oncologist gave me valuable suggestions. He directed me to support groups, where I found good answers to the many questions I had about my risks and options. I researched my options and, based on my BRCA risk, decided to have a bilateral mastectomy with immediate reconstruction. I selected two amazing plastic surgeons to perform DIEP, an advanced reconstruction procedure that, without implants, leaves a woman's breasts whole, made of my own soft, warm, living tissue. I do not feel as if I lost my breasts...only my risk of breast cancer.

My breast surgeon said to consider myself cured. I still have regular breast exams, although my chance of having breast cancer is now 1 to 2 percent, which is lower than the general population's 8 to 12 percent risk. I am satisfied that I have now done all I can to prevent cancer.

Looking back over my family's history, I am reminded that we are a tough bunch—survivors and co-survivors all. Thanks to medical research advances, the future of my children, and all BRCA mutation carriers is bright.

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 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 retested. The second test also came back positive. Without a doubt, Miguel had this disorder; though he still seemed completely healthy. The doctor told us that children with this rare genetic disorder are unable to break down excessive protein and 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 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, 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, and Boy Scouts. Since 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 had 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 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, cirrhosis of the liver, 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 (a procedure similar to donating blood) to keep the iron from accumulating in my body and damaging my organs. 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 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, despite drinking plenty of water and other beverages. In spite of my increased drinking habits and normal appetite, I somehow lost 40 pounds. Finally, my discomfort from the skin condition became so severe that I went to the emergency room, where I was diagnosed with Type II diabetes. Apparently, I had actually had this condition for some time.

Since my diagnosis, I have learned a lot about my family and 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 realized my risk was greater than that of someone without a family history.

Additionally, 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. Also, 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.