Code Talkers

***Genome* readers honor genetic counselors who interpret complexity with compassion.**

SARAH

Sarah Ruppert
Life Changer

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Nominated by
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Photographs by
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Sarah Ruppert by Erica Stoecker

I would like to nominate Sarah Ruppert for the Code Talker award. She combines the compassion and expertise needed not only to fully comprehend the science behind genetic testing but also to translate that knowledge into crucial, understandable information for her clients.

After receiving negative test results for the BRCA mutations when I was diagnosed with breast cancer at age 40 in 2010, I felt like a genetic testing veteran. My oncologist's recommendation to see a genetic counselor in 2014 was not well received. Why did I need counseling? Going to that counseling appointment turned out to be one of the best decisions of my life. My genetic counselor, Sarah Ruppert, identified the right test, saved me from an impending second cancer, and enabled my family members and me to take action to prevent additional cancers. From first meeting to communicating results, through screenings and treatments, Sarah's clarity, empathy, and support were beacons of light in a time of emotional and medical chaos.

My initial meeting with Sarah was extremely informative. She walked me through my family history, the tests she was recommending, and possible outcomes. I was amazed by the complexity of the science behind genetic testing, and even more amazed by her ability to communicate the theory and the practical impact in a manner understandable to me. In the end, she selected a panel of tests based on my individual and family history.

A few weeks later, Sarah gave me the results. A pathogenic mutation had been identified in my MSH6 gene, which increases the risk of Lynch syndrome. Although Sarah had prepared me to hear that my grandfather's colon cancer could be related to Lynch, and we were testing for Lynch genes, learning that I was at high risk of developing multiple other cancers was still an emotional shock. However, Sarah delivered the news with the perfect balance of emotional gentleness and factual professionalism. My husband, my sister, and I then visited Sarah, and she patiently and thoroughly reviewed with us the risks associated with Lynch, recommended screenings and medical tests, and answered all our questions. My sister, and later my mother, were tested, and we all have the mutation.

Lynch syndrome results in an increased risk of several cancers, notably bowel, womb, and ovarian. Unbeknownst to me, my womb was already on the road to cancer. Upon receiving the Lynch diagnosis, my oncologist immediately scheduled a biopsy, which identified complex atypical hyperplasia, a precancerous womb condition that, in a normal population, leads to cancer approximately 30 percent of the time. My mutation meant I had a greater than 60 percent chance of womb cancer at a median age of 46, and I was 45. Additionally, the tamoxifen I had been taking for my breast cancer increased the risk of womb cancer. It was the perfect storm. Without the benefit of the genetic knowledge, I would not have discovered it until later, maybe too late. I met with a gynecological oncologist. Four days later, she removed my womb and ovaries.

Sarah also advised me of the need for a colonoscopy, which, under traditional screening recommendations, would not have occurred for several more years. A flat polyp, quite possibly on its way to becoming cancer, was removed, and additional polyps were removed during a second colonoscopy eight months later. Once again, learning of the genetic condition enabled me to avoid cancer.

The benefit extended to my family. Sarah assisted my sister and mother in deciding to be tested and helped them understand the implications of their positive results. They both had risk-reducing surgeries and prompt colonoscopies.

The results also are furthering our understanding of the relationship between Lynch syndrome and breast cancer, an area of medical uncertainty. After my genetic diagnosis, my oncologist had my breast cancer tissue tested. It showed a lack of expression of the MSH6 protein. I have shared this information with several research studies.

The information learned through genetic testing has empowered my family and me to make decisions and take action to make us safer from an early, cancer-caused demise. I am eternally grateful for the incredible compassion and competence with which Sarah guided me through this life-changing journey.

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SARAH, ERICA