TRACKing Rare Mutations

After being treated for thyroid cancer with a rare mutation, Susan is helping educate others.

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Susan, a pennsylvania resident in her 50s, was diagnosed with thyroid cancer in 2004. She underwent surgery to remove the gland, followed by standard treatment with radioactive iodine to destroy any remaining cancer cells. (Susan asked that we not use her last name.)

“I was told not to worry about it; I had a 98% chance of a cure,” she says.

But in 2008, she had a recurrence. She went to a cancer center in Philadelphia for further evaluation. A tumor was found on her spine, which was treated with radiation. Then scans revealed that the cancer had spread to her lungs.

Over the next decade, as her cancer progressed, Susan received more radioactive iodine treatments at very high doses. She developed a tumor in her neck, which was removed and grew back twice.

Running out of options, in November 2017, she saw a new oncologist who suggested genomic testing. “I said ‘Yes, sign me up!’” she recalls. “I was concerned about the financial aspects because some insurance companies don’t pay for it, but at this point, I was desperate and said, ‘Let’s do it.’ It ended up being one of the best appointments ever.”

The tests revealed that Susan’s cancer had a rare DNA mutation, known as an NTRK fusion, that spurs cancer growth. These gene fusions occur in only around 1% of cancers overall, but they’re more common in certain rare malignancies, incuding some types of thyroid cancer. She also learned that there was a clinical trial of an experimental drug that targets this specific genetic alteration. In August 2018, she started taking Vitrakvi (larotrectinib), a twice-daily pill. The treatment was approved in November that year.

“Within five days, my neck tumor was literally smoother where the bump was,” Susan says. “My wheezing when I exercised went away. My scans showed tumor shrinkage right away.” By January 2019, her scans showed a 50% reduction, and her cancer has been stable or getting smaller ever since.

Susan searched Facebook and other social media and found about a dozen other patients taking the same drug. She started messaging them, and they all replied within a day. “They said ‘I’m so

glad you found me,'" she recalls. "People wanted to connect with others who had this thing that's so rare."

Through her advocacy, Susan met Jim Palma, executive director of the TargetCancer Foundation, and joined the advisory council for the TRACK trial, providing a patient perspective and helping design the study. TRACK (supported by Vitrakvi manufacturer Bayer) aims to enroll 400 participants with rare cancers who will receive genomic testing to see whether they have tumor mutations that can be matched to targeted therapies and whether these treatments delay disease progression.

"Genomic biomarker testing has to be made more affordable and accessible," Susan says. "It's a shame how many people we've lost because they weren't tested. Everyone should have access. It's a game-changer. It can completely change the course of your cancer journey."
