



2022 Project Brain Light Essay Contest *Grand Essayist*

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Biomedical Research and My Life

A mysterious creature emerges. Infinite potential. Always growing. Always reproducing. What once seemed normal is now completely out of control. This mysterious creature turns out to be a disease called cancer. In 2015 my mother had an unresolved pain in her jaw and after two months of various doctor appointments, she was diagnosed with thyroid cancer. It was a stressful and worrisome time, the hospital visits, the surgeries, and the radiation treatment, but my mom got through it. Thyroid cancer is like other cancers, where cells reproduce too much and develop into a tumor. The most common treatment for this type of cancer is surgery to remove the thyroid glands. However, the thyroid is an important organ that releases hormones to control the metabolism and other vital functions of human body. Without the thyroid, people need to take synthetic thyroid hormone to maintain and sustain those functions. Thus, patients with thyroid cancer must rely on synthetic hormones for the rest of their lives after their thyroid gland is removed.

The process of diagnosing thyroid cancer starts with a procedure called fine-needle aspiration biopsy. Doctors take a small sample from the thyroid nodules and send it to a pathology lab to look for cancerous cells. However, the results of the biopsy can be ambiguous and indeterminate. Over one-third of biopsies are indeterminate. Thus, many doctors choose to remove a patient's thyroid even when it might not be cancerous in a conservative approach. As described above, many patients would have to take thyroid hormones for the rest of their lives. Fortunately, in recent years, the advance of biotechnology offers a solution to this problem. Biological and medical research has shown that many types of cancer harbor mutations in the K-Ras gene. The K-Ras gene is a gene that responds to cell signals and sends responses to instruct the nucleus to multiply. If a K-Ras gene mutates and continues to tell a cell to replicate that can cause excessive and uncontrollable cell growth. Out-of-control cell growth is the hallmark of cancer. Based on those findings, scientists realized that doctors could use the mutations in the K-Ras gene to help identify patients who would be at high risk for developing or having thyroid cancer and should require surgery to remove the thyroid glands.

How did scientists know the "arrangements of bases" of a gene? It all began when Fredrick Sanger developed the DNA sequencing method in 1977, he paved a revolutionary way to not only find the normal sequences of a gene but also detect the "variant" ones. Some of these variant sequences have mutations that alter the functions of genes. Following the invention of automation of the Sanger sequencing process, scientists around the world could sequence many genes and discover mutations in organism's genome. With automatic sequencers, the Human Genome Project successfully sequenced 99.9% of the human genome in 2003. After this historical breakthrough, continued innovations in DNA sequencing methods have lowered the cost and shortened the time for obtaining the gene sequences. All these scientific advances make the sequencing of cancer genomes possible, and physicians can use the information from the sequencing to make informed decisions for their patients. As matter of

the fact, my mother's doctor sent her thyroid biopsy sample for sequencing and found she had Ras gene mutations. Because of the confirmation of the Ras gene mutations, she had a total thyroidectomy, and her life was saved by biomedical research on DNA sequencing.

Today, genomic screening is widely used to correctly identify cancer. Specifically, in thyroid cancer, a test called ThyroSeq is commonly used. This test measures 112 genes and changes that are linked to thyroid cancer, such as mutations, gene fusions, changes in copy number, and/or gene expression. Combining biopsy results with the information from the sequencing test, doctors can make a more accurate diagnosis and personalize the treatment plans for patients. My mother has benefited from those types of biomedical research in that her thyroid nodules were identified as positive in a pathology report with confirmation from cancer gene sequencing. As a result, I am grateful for the advances in DNA sequencing that saved my family from the despair of cancer.

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