Life On The Edge of Independence: My Brother and Tuberous Sclerosis

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Rounding the corner of Powhatan Street onto Kirby Road, my mother sweeps her head side to side checking for cars approaching; my novice driving skills frighten her. My older brother sits in the back of the car, his head tilted up, listening to Frank Sinatra with his phone next to his ear. After completing my left turn, I accelerate up and down the rolling hills of Kirby. At the end of a strait away, I spot a red stop light and I begin to press down on the brake pedal. My mother turns her head to the back of the car to check my brother; he looks out the window with his mouth open, mumbling a few words of the Frank Sinatra song. A few more turns in the hills of Northern Virginia and we have arrived at our destination: Safeway grocery store.

I pull up into a space close to the store, and put the car in park. My mother instructs my brother to pick up his things: a lunchbox, a mask, and a name tag. We all exit the car, and walk up to the sliding-door entrance. My mother continues to give my brother instructions and reminds him of all his tasks of a courtesy clerk. Once my brother has been escorted to the sign-in station, my mother and I turn to our shopping duties. Realizing that my brother is out of our hands, we can breathe easier.

My mother and I stroll around the store and check off a list of items we need. Because we come to the store so often to drop off my brother for work, we know exactly where to find all of our items. We collect frozen meals for dinner, some cheap cereals for breakfast, and some snacks for my brother.

Walking down the frozen aisle with my mother, I pass the various brands of ice cream: Breyer's, Ben & Jerry's, Haagen-Dazs, Edy's, and more. A teenager dressed in black with his Safeway badge pushes a cart around the corner of the aisle: a courtesy clerk like my brother. He greets me with an open-mouth smile as he pushes his glasses up on his face. He leans down into his cart, grabs a box of frozen vegetables and opens the door to the freezer. The freezer makes

the usual loud humming sound, amplified by the open door. The worker sets some turned down items from the cash register back in their appropriate positions inside the freezer. His cart empty, he pulls his arm out from the freezer, which claps back into place with a loud slam. Just as he finishes, my brother walks around the corner.

As my brother approaches the other courtesy clerk, they form a perfect juxtaposition: a normal teenager standing next to a flawed one. My brother's differences are quite clear next to someone like him. My brother hunches his back over while the other teenager stands straight. My brother has his hair jumbled while the other teenager has his combed. My brother's glasses are smudged with fingerprints while the other teenager's glasses are clean. My brother strolls around the Safeway until he is told what to do while the other teenager returns items independently. My brother had to be driven to his job at age eighteen while the other teenager drove himself to the same job. They both grew up in the same neighborhood, they both went to the same elementary school, and they both work the same job but are still so different in their abilities. Why?

The story begins in 2004 in Tampa, Florida. In a small doctor's office only a few blocks away from Hillsborough Bay, my brother was taken at the ripe old age of two to see my grandfather at his dermatology practice. The building of gray concrete surrounded by moss-covered trees would be a place I would visit after I was born. My grandfather had routinely examined my father when he visited; because my father grew up in Florida, he occasionally had a piece of skin that had to get checked out. My parents simply brought my brother along with them to their routine visit to my grandparents. After taking a look at my father, my grandfather had just enough time to examine my brother before his next appointment. A bumpy patch on my brother's lower back, believed to be a birthmark by my parents, would change our family's life.

The bumpy patch, also known as a shagreen patch, indicated the presence of Tuberous sclerosis Complex (TSC) in my brother, a genetic disease which causes benign tumors to form in various vital organs such as the brain, kidneys, heart, lungs, eyes, and skin. Tuberous Sclerosis Complex appears in two types: type 1 and type 2. My brother has type 1, corresponding to a mutation of the TSC1 gene located on the ninth chromosome. The TSC1 gene instructs how to create a protein called hamartin which helps regulate cell growth and size. The protein limits the proliferation of a protein called the mechanistic target of rapamycin (mTOR), which helps with cell growth and repairs. When the mTOR protein is let loose by a mutation of hamartin, enlarged and abnormal cells form.[1]These enlarged cells, typically found in the brain in TSC brain lesions, cause mental issues that separate my brother from other teenagers his age. My grandfather would explain to me at a young age that my brother's brain simply is wired differently than other people. When my grandfather first used this terminology, I did not see the effects of the rewire.

Ever since his diagnosis, my brother has travelled to Boston every year to visit a specialist on TSC. I would usually stay at home with a friend while my parents took my brother. However, one year I travelled to Boston to meet the specialist and spend a few days exploring the city. During my trip, my parents underwent several tests along with my brother to learn about how my brother may have acquired the disease. TSC occurs when the TSC1 mutation spontaneously forms or when the mutation is inherited. Most TSC cases originate from a spontaneous TSC1 mutation, where a mutation randomly occurs in a newborn child. TSC is inherited when one parent of a child has TSC, even if the parent was unaware they had the disease. The children of TSC patients often have completely different symptoms than their

[1] National Center for Biotechnology Information. (2021). *TSC1 TSC complex subunit 1 [Homo sapiens (human)] - Gene - NCBI*. Nih.gov. https://www.ncbi.nlm.nih.gov/gene?Db=gene&Cmd=ShowDetailView&TermToSearch=7248

parents; a parent may never be diagnosed with TSC because they have never shown symptoms, but their child may have severe symptoms of the disease.[2] The testing on the Boston trip determined that my brother had acquired the disease spontaneously. (Office of Communications and Public Liaison National Institute of Neurological Disorders and Stroke, 2020)

Alongside the annual Boston trips, my brother requires scanning of his kidneys. TSC patients often develop kidney problems in two different types: cysts and angiomyolipoma. Benign growths of muscle cells and fatty tissue, cysts can cause kidney issues. In a few cases of TSC, childhood cysts lead to bleeding, anemia, and kidney failure. Angiomyolipomas are a more common type of benign growth in both kidneys and typically produce no symptoms. In a few rare cases, Angiomyolipomas grow large enough to cause pain in TSC patients and even result in kidney failure. Angiomyolipomas occasionally bleed, which causes pain in most patients. If this bleeding is severe and the blood does not clot naturally, patients experience dramatic losses in blood pressure and require urgent medical attention.[3] While my family has not been threatened with kidney complications, my brother will require close kidney observation for the rest of his life.

After one Boston trip, my brother returned with a white cloth wrapped around his head and wire attached to a hand held monitor. My parents explained to me that the cloth was used to monitor my brother's brain activity for seizures. TSC patients, although mostly with TSC type 2, experience seizures starting at a young age. Three types of seizures plague TSC victims. Infantile spasms appear in the earliest stages of life; babies ranging from 4 to 8 months experience this

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[2] MedlinePlus Genetics. (2011). *Tuberous sclerosis complex: MedlinePlus Genetics*. Medlineplus.gov. https://medlineplus.gov/genetics/condition/tuberous-sclerosis-complex/

[3] Overwater, I., Rietman, A., van Eeghen, A., & de Wit, M. (2019). Everolimus for the treatment of refractory seizures associated with tuberous sclerosis complex (TSC): current perspectives. *Therapeutics and Clinical Risk Management*, *Volume 15*, 951–955. https://doi.org/10.2147/tcrm.s145630

type of spasm. Infantile spasms occur when babies are going to bed or are waking up. Babies that experience these spasms may jut their arm out uncontrollably or flex muscles.[4] Partial seizures are the most noticeable type of seizure to the average passerby. One might notice a partial seizure when a TSC teenager or adult jerks their head, repeatedly move their arms, or lock their eyes upward. Absence seizures also appear in TSC patients, where they decrease their response to other people. Sometimes my brother will stare off into the distance with his mouth open, which I believe may be an absence seizure. Although ninety percent of TSC patients have epilepsy, few antiepileptic drugs benefit TSC patients.[5]

The symptoms of TSC change the lives of everyone around them. Whether a brief flex of an extremity or a kidney failure or an outburst of emotion, TSC patients often embarrass their family members. For me, the embarrassment progressed with age.

At a young age, I did not notice any differences between my brother and the kids around him. When I was in Kindergarten, my grandparents would take my brother and me to Whole Foods in Lyon Village outside of Washington D.C. to buy some cookies. I remember telling my brother that "I could smell the cookies already" even though we were a few blocks away from the store. My brother, likely having an absence spasm, did not respond to my comment and simply stood still. My friends sometimes ignored me, so I figured that my brother had just done the same. At the time, my brother appeared the same as everyone else around him. His

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[4] Office of Communications and Public Liaison, National Institute of Neurological Disorders and Stroke. (2017). *Tuberous Sclerosis Complex (TSC)*. Epilepsy Foundation. https://www.epilepsy.com/learn/epilepsy-due-specific-causes/structural-causes-epilepsy/specific-structural-epilepsies/tuberous-sclerosis-complex-tsc

[5] Office of Communications and Public Liaison National Institute of Neurological Disorders and Stroke. (2020). *Tuberous Sclerosis Fact Sheet | National Institute of Neurological Disorders and Stroke*. Nih.gov. https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Tuberous-Sclerosis-Fact-Sheet

handwriting was horrible, and my handwriting was horrible. My brother had no more than five or six friends, and I had no more than five or six friends. My brother rode the bus to and from school, and I rode the bus to and from school. My brother's Artwork was simple and abstract, and my Artwork was simple and abstract.

Figure 1

My Childhood Artwork vs. My Brother's Childhood Artwork

Note. This is a comparison between my artwork (right) and my brother's artwork (left) that we both created age 10.

However, as we aged, the differences between my brother and I emerged. Most differing features emerged slowly, but the most obvious was performance in school. I had stayed consistent through most of my middle school and high school career, performing well. My brother completed his early education well, but his grades and performance declined in middle school and high school. My family began to support him in school, helping him complete homework and study for tests. Our physical differences arrived next. My brother developed

spinal issues, as he hunches often over when sitting at his desk or in his room. By high school, my brother and I were about the same height even though he was taller most of his life. Today, I stand about 3 inches taller than my brother. Perhaps the most obvious are simple every day capabilities. I am in the process of earning my drivers license, while my brother has never driven a car. My brother has to be monitored while he does his homework, while I do my homework on my own. My brother plans to stay an extra year in high school while I plan to go straight to college.

Figure 2

My Teenage Artwork vs. My Brother's Teenage Artwork

Note. I created the landscape on the left in freshman year of high school and the painting on the right was created by my brother in sophomore year of high school.

July 26, 2018, Dallas, Texas. My mother, brother, and I exited a taxi and entered the Hilton Anatole. The lobby contained several banners, each reading "TSC Global Conference." in blue, orange, and green letters. We arrived around 7:00 pm just before sunset and most of the conference's activities were over. We took an elevator up to our hotel room and saw a few parents watching their children run around in the hallway. At 8:00, we took an elevator down to

the lobby floor and walked over to a large ballroom for dinner. The TSC Alliance prepared a large buffet for the conference attendees and assigned each attendee to a table around the room with random others. I put my plate down last out of those seated at my table: all of whom appeared to be adults with research experience. I greeted them with a smile, and they invited me to sit with them. Then someone started the usual icebreaker; what is your name and why are you here? My tablemates discussed their researching responsibilities, most of which I did not understand. When the chain of introductions arrived at me, I explained that I am the sibling of a TSC patient here to learn more about the disease. The woman next to me, probably in her sixties, discussed herself. I expected more research jargon, but instead she explained that her son had had TSC for almost 25 years. She described her son's childhood, his experience with bullying, his struggles with college and career. She talked about kidney scans, endless appointments, and rising bills. These challenges had clearly worn down on the woman, who spoke in a quiet and pessimistic voice. Everyone else at the table looked down to reflect on their challenges with the disease. The researchers may have thought about the TSC patients they had met in the past, but I took an honest look at my brother.

I tried to imagine my brother a decade older. I couldn't do it. My family had spent so much time on my brother and I couldn't think of him becoming an adult. The woman at my table explained that her son still lived with her. But I could not think of my brother living with my parents either: "he is capable of living on his own isn't he?"

Since that woman turned my eyes on my brother's future, it has become increasingly hazy. My brother can work at Safeway, but my mother and I still need to guide him. My brother can still go to high school but my family still needs to help him study. My brother lies right on the edge of independence.

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