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An Introduction to Moyamoya Disease

Michael Shen

Stephanie, a good friend of mine, experienced her first migraine on her twelfth birthday. From then on, she had migraines at least once a month, sometimes with additional symptoms such as a partial loss of vision, as well as nausea and numbness. In high school, she was an active girl and an ace on the tennis court. I remember that her headaches were sometimes so debilitating that she missed class and fainted once during a varsity game. Her mother, May, often consulted her family doctors about her migraines. However, despite her more concerning episodes, the doctors never suspected an underlying cause.

Looking back, May tells me she wishes she had listened to her motherly instincts and had urged more adamantly for a brain scan, which the doctors deemed unnecessary given that Stephanie only had a few other symptoms and that she had recovered from her spells after just a few hours of sleep. Stephanie remembers being reassured that migraines like hers were not too uncommon for teenage girls.

In September of her senior year of college, Stephanie went on vacation to Las Vegas. I had kept in touch with her since high school, but it had been a month since we last talked. I did not know that it would be another month before I heard from her again. A week prior to her vacation, she had experienced an unusual episode in which she had awoken unable to move her neck. Concerned, May had taken her to the doctor, but no conclusions were made that day. Stephanie later found out that she had suffered a transient ischemic attack (TIA), frequently referred to as a “mini-stroke”. On her last day in Las Vegas, Stephanie began to feel an oncoming migraine, which was exacerbated by the smoke-filled lobby of her hotel. She collapsed and was taken to the hospital in a dire condition.

At the hospital, it was determined that she had suffered a hemorrhagic stroke, and her parents were notified immediately. They were given an extremely grave prognosis and asked to make a choice between two equally unbearable ends: the first was to let their daughter die peacefully; the second was to drill a hole into her skull and induce a coma. They declined medical intervention and prayed by Stephanie’s bedside. As May recalls, her daughter’s brain was swollen and under great pressure inside the skull; it wouldn’t be long before she was brain-dead. However, when the doctor arrived for his final round, he noticed signs that prompted a reevaluation of her mental status. It seemed like a miracle—Stephanie’s body refused to succumb to death.

At this point, the necessary brain scans and angiogram were ordered, and the evidence viewed. Speculations about Stephanie’s condition began to surface in the minds of those treating her. Moyamoya disease (MMD) is a rare and relatively unknown disorder. Her doctors had heard of it but had never

seen it, nor were they sure she even had it. In fact, I doubt I will ever come across this disease in my studies as a medical student. Moyamoya is characterized by the progressive occlusion of both internal carotid arteries due to excessive growth of smooth muscle cells and luminal blood clot formation.^{1,2} As a compensatory reaction to the lack of blood flow to the brain, numerous tiny and tortuous collateral arteries are formed. These are often thin, dilated, and subjected to high flow stress, increasing the risk of microaneurysms.^{1,2} Patients often present with cerebrovascular problems such as TIAs, ischemic stroke, intracranial hemorrhage, migraines, or seizures.

The disease was first described in Japan, and its appearance on diagnostic angiograms was likened to a “puff of smoke,” or “moyamoya”, in Japanese.² This telling arrangement of tiny blood vessels was revealed in Stephanie’s angiogram.

An analysis of patients in the western United States revealed an overall prevalence of 0.086 per 100,000.¹ However, a four-fold greater incidence was found in Asian-Americans than in Caucasian Americans, supporting the possibility of a genetic etiology. Stephanie, who is Chinese-American, fits this higher-risk profile. It has also been observed that the incidence of this disease is almost two times higher in females as in males. Furthermore, the incidence of the disease peaks in children around the age of 5, as well as in adults in their mid-40s.^{1,2}

As is the case with many rare diseases, proper diagnosis is often hindered by many factors. Because Moyamoya is relatively unknown, it is often misdiagnosed as multiple sclerosis or simply as migraines, as in Stephanie’s case.³ A delay in diagnosis can mean a delay in proper treatment and ultimately a higher risk of life-changing implications from stroke. The situation is made worse when doctors attempt to treat the symptoms rather than work toward unveiling the underlying cause.

There is no cure for Moyamoya, but current treatments are successful in reducing the risk of future strokes by surgical revascularization of the cerebral hemispheres.¹ Stephanie was lucky to be treated at Stanford University Medical Center, one of the few places in the country that specializes in Moyamoya.

The first time I talked to Stephanie after her Las Vegas incident was during her recovery from surgery. I spoke to her as I would have on any other day, listening as she stumbled on words and repeated thoughts that she had voiced just minutes before.

May once told me her greatest fear was that when her daughter came back, she would not be the same Stephanie. It was a strangely emotional experience for me, knowing that she



would most likely live free of risk after surgery, yet wondering if her mother's fears could somehow prevail.

Today, the most difficult challenges of Moyamoya seem to be behind her. Stephanie has had no cognitive or motor impairments as a result of the stroke and brain surgery. She has bounced back magnificently. From my conversations with Stephanie and her family, I feel that the events of the past year have laid upon them a new lens through which they view their existence. Having experienced the darkest and brightest moments of death and of life, they now live with a secret spirituality that I do not know but can only presume. And there is nothing more that I can be or feel, than thankful for Stephanie's full recovery—personality and all.

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