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VIEWPOINT

VOICES OF CARDIOLOGY

Has Your Physician Heard of Williams-Beuren Syndrome?

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As a child, my family would come together during the holidays, and the grandchildren would play in my grandmother's basement. However, every time we asked our cousin Cynthia (her name has been changed to protect her privacy) to join us, she would have difficulty participating. As she aged, she began to have regular admissions to the hospital, but at the time, we did not understand what for. For almost 4 decades, Cynthia was undiagnosed, until a chance encounter with a cardiologist.

During my senior year of high school, Cynthia was admitted to an inpatient ward for complications of her atrial septal defect and mitral regurgitation. A young cardiologist was doing his bedside rounds with Cynthia's roommate when he glanced over at Cynthia finishing her breakfast. Before departing the room, he approached my family and said, "This is my first time seeing Williams-Beuren syndrome."

The family was baffled. This was the first time a diagnosis was provided to us. Williams-Beuren syndrome is a multisystem genetic disorder caused by deletion spanning from 1.5 to 1.8 million base pairs, involving 26 to 28 genes (1). The syndrome is seen in approximately 1 in 10,000 births and is characterized as a microdeletion disorder or contiguous gene deletion disorder (1).

The syndrome was first described in 1961 by Dr. John Cyprian Phipps Williams (2). He described patients who had cardiovascular abnormalities, mental "subnormality," and growth retardation (2). Cynthia was born in 1965, leaving the diagnosis essentially unheard of at the time. This young cardiologist diagnosed this condition instantaneously based on the unique facial features of the patient, which are classically described as "elfin face," referring to a wide mouth with long phyltrum and thick lips (3).

A wide ranges of cardiovascular abnormalities are typically seen. Supravalvular aortic stenosis has been described in approximately 70% of Williams-Beuren syndrome patients (4). There is stenosis of large and medium sized arteries due to thickening of vascular media, which is derived from smooth muscle overgrowth (1). The arterial narrowing can be diffuse, involving the descending aorta, aortic arch, and mesenteric and intracranial arteries. In certain cases, the arterial disease can be isolated (1). In all cases, there is an increased carotid artery intima media thickness related to a generalized elastin arteriopathy (1). Ventricular and atrial septal defects are uncommon. Myxomatous degeneration of mitral or aortic valve leaflets can be found in approximately 20% of patients (1). Hypertensive disorder usually begins in childhood and is seen in approximately one-half of all cases, although the cause of hypertension is not completely understood (1). The major causes of death in these patients are cardiovascular complications (1).

I reflect on Cynthia's hospital encounter and feel great admiration for this physician cardiologist. He not only treated patients but also brought clarity to medical issues and challenges that patients as well as family members face. I wanted to become the voice of reason and clarity. It became engraved in my mind that no matter how many years it took, no matter how many obstacles or rejections I faced along the way, it was my calling in life to become a cardiologist. I felt a

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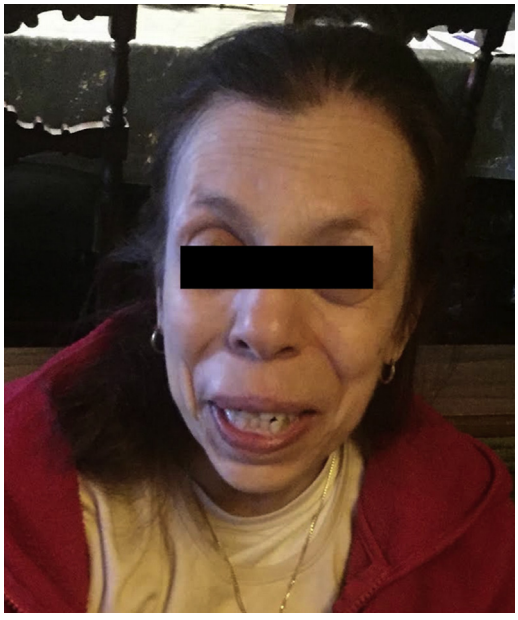
FIGURE 1 Elfin Face, Long Phyltrum, and Thick Lips



proud sense of inherited duties and responsibilities had called upon me to move forward in a career in medicine as a cardiologist.

This journey took me from Port Richmond High School in New York City, a public school with an approximate graduation rate of 50%, to obtaining a bachelor's degree in Biomedical Science. Subsequently, I attended medical school in Guadalajara, Mexico, where I immersed myself in a new language and culture while studying medicine.

I am currently a second year resident physician in Internal Medicine, completing my training close to home in New Jersey. It is during my encounters daily that I am reminded of the moment of Cynthia's diagnosis. I come to work each day with motivation to acquire as much knowledge as I can, to immerse myself in numerous projects to contribute to the improvement of the health care system, and to the advancement of the field of cardiology. As I prepare my upcoming cardiology fellowship application, I reflect on what got me to this point, and I smile. To this day I love sharing my story with fellow residents and with my teaching attendings. To my surprise, including the most senior, not one physician I have worked with has encountered a patient with Williams-Beuren syndrome.

I would like to honor Cynthia (**Figure 1**), increase the awareness of her disorder, and share how a small moment from a physician in the medical profession can alter the life of young adults. I would like to thank the influence of that cardiologist who inspired a lost 17-year-old to find a renewed sense of purpose.

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