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Health & Medicine

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Williams Syndrome is a genetic pediatric illness that was first observed in New Zealand by the reclusive Dr. J. C. P. Williams while working as a Registrar at Auckland's Greenlane Hospital in 1961. According to Physiopedia (2011), Dr. Williams noted that 4 of his patients had; “ an association between supra valvular aortic stenosis and the common physical and mental characteristics of this patient population ... it “ may constitute a previously unrecognized syndrome”. After some time, A. J. Beuren came across his own set of 11 patients who also had the same symptoms as described by Dr. Williams in the past. Thanks to his contribution to the study of what was then an unknown illness, the disease was named after the two men who contributed the most its understanding, Williams-Beuren Syndrome. However, the illness has also come to be known by other illnesses over the years. Physiopedia (2011) lists the other names as follows:

Williams-Beuren Syndrome

Beuren Syndrome

Elfin Facies Syndrome

Elfin Facies with hypercalcemia

Hypercalcemia-Supravalvar Aortic Stenosis

Infantile hypercalcemia

Supravalvar aortic stenosis syndrome

WBS

WMS

WS

Dr. Aneal Khan M. D., who authored an article regarding Williams Syndrome for Medscape Reference (2011) described the clinical manifestation of the illness to include; “ a distinct facial appearance, cardiovascular anomalies that may be present at birth or may develop later in life, idiopathic hypercalcemia, and a characteristic neurodevelopmental and behavioral profile”. The disease does not seem to afflict any one specific gender, ethnicity, race, or socioeconomic background. However, it does seem to be most prevalent among children, affecting 7500 of every 20000 births. The most common symptoms of the birth defect according to Dr. Rob Hicks (2011) include but are not limited to:

Upturned nose

Widely spaced eyes

Wide mouth with full lips

Small chin

Slightly puffy cheeks

Irregular, widely spaced teeth

Various in depth studies of the illness link Williams Syndrome to the genetic make up of an individual. Dr. Rob Hicks (2011) , author of “ Williams Syndrome” for BBC Health explained that WS afflicted individuals most often have a defect in the DNA chain of chromosome 7 with about 26 genes deleted from its long arm. Unfortunately, chromosome 7 is one of the most important elements of the human gene as this particular gene is tasked with the manufacture of the protein elastin which according to Dr. Hicks (2011), is “ is responsible for providing strength and elasticity to blood vessel walls. “ Babies born with Williams Syndrome are often underweight upon birth with difficulty in gaining weight and have certain development defects such as

speech problems with the ability to speak not becoming present until the age of 3. However, the most concerning matter of this illness is the widespread Cardiovascular complicationsassociated with it. The lack of development of elastin in Chromosome 7 prevents the manufacture and development and elastin within the child, weakening the cardiovascular walls of the child and presenting him with various cardiovascular illnesses over time. (Williams Syndrome Association, 2011). A WS afflicted child faces major health struggles, with the cardiovascular illnesses proving to be life-threatening. The Williams Syndrome Association explains that these heart and blood vessel problems (2011) tend to be highly complicated because :

... majority of individuals with Williams syndrome have some type of heart or blood vessel problem. Typically, there is narrowing in the aorta (producing supravalvular aortic stenos is SVAS), or narrowing in the pulmonary arteries. There is a broad range in the degree of narrowing, ranging from trivial to severe (requiring surgical correction of the defect). Since there is an increased risk for development of blood vessel narrowing or high blood pressure over time, periodic monitoring of cardiac status is necessary.

WS symptoms control procedures suggested include avoiding extra calcium and Vitamin D, physical therapy, speech therapy, and other treatments depending upon the present symptoms of the illness. A child afflicted with WS will do best under the care of a geneticist with experience in handling Williams Syndrome cases. In the end, about 75 percent of those afflicted will have some sort of mental retardation and require long term professional care without living any semblance of a normal life. (New York Times: Health - Williams Syndrome, 2009). The U. S. National Library of Medicine A. D. A. M. reference (2011) explains that the illness can only be controlled at present,

no cure is imminent.

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