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Facial Dysmorphism with Precordial Systolic Murmur of a Young Boy Williams Syndrome

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Abstract

A 7 years old boy diagnosed Williams's syndrome by clinically with the help of some investigations. This child having mal-occlusion of multiple teeth with dental carries, large forehead, small chin, puffiness around both eye, looks like-elfin facies with low IQ. He has a systolic murmur (Grade-4/6) in the base of the heart. Echo, Doppler gradient found in the just supra-valvular area, 128 mmHg (Supra- valvular Aortic stenosis) and MVP (Mitra valve Prolapse). This child also having blockage of naso-lacrimal duct, causing watering of eye. William's Syndrome (WS) is a neurodevelopmental, multisystem genetic disorder characterized by distinctive personality traits, facial dysmorphisom (elfin face‖) and congenital cardiac defects, of which supravalvular aortic stenosis is the most common lesion found. It is characterized by Congenital Heart Defects (CHD), Skeletal and renal anomalies, cognitive disorder, social personality disorder and dysmorphic facies. WS is a rare familial multisystem disorder occurring in 1 per 20,000 live births.

Introduction

William's Syndrome (WS) is a rare but well recognized neurodevelopmental disease affecting the connective tissue and the central nervous system. It also affects cardiovascular system and eye. The syndrome was first described in 1961 by Newzealander JCP Williams [1,2]. It is caused by deletion of about 26 genes from the long arm of chromosome 7 and it occurs 1 in 7,500 to 1 in 20,000 live birth [3,4]. The underlying molecular mechanism is a submicroscopic chromosomal deletion involving the elastin gene (ELN) at 7q11.23 [5]. It is present at birth and affects boys and girls equally [3]. WS having infantile hypercalcamia, hoarseness of voice, hyperacusis, endocrine abnormalities, growth retardation, orthopedic problems and renal abnormalities may also be associated [6,7]. Due to variability in the clinical findings, diagnosis is usually made during mid-childhood when the characteristics facial features, cognitive profile and cardiac findings become more apparent [8]. Beuren in 1964 independently described the syndrome noting also typical dental anomalies [9].

Case Presentation

A 7-years old boy diagnosed Williams Syndrome by clinically with the help of some investigations. This boy actually referred from eye hospital for preoperative (Dacrocystorhinostomy/DCR operation) cardiac consultation. The child was having large forehead, small chin, puffiness around the both eyes, nose upturned, long philtrum looks like-elfin facies‖ associated with malocclusion of multiple teeth with Dental carries. He was of low IQ as he was in the same class for 3 years with stunted growth, height 125 cm, weight 22 Kg. Precordial examination revealed systolic thrill and systolic murmur (Grade-4/6) in the base of the heart.

His past history was not contributory except for few episodes of respiratory tract infections since childhood. In the other family members, evidence of congenital anomaly or mental retardation was not found. According to primary symptoms, clinical finding such as dysmorphic facies, cognitive disorder and congenital supravalvular aortic stenosis, dental mal-occlusion and eye problem (Lacrimal duct not patent), WS was the first diagnosis.

Chest X-ray revealed cardiomegaly, [10] leads ECG shows normal findings. Echocardiogram showed normal chamber size, normal aortic valve leaflet but supravalvular aortic stenosis due to narrowing of ascending aorta (PPG128mmHg) & prolapse of anterior mitral leaflet , other valvular structure and pulmonary artery were normal. Other laboratory parameter, like Hb%, ESR, Serum Calcium level was normal. We suggest a more attention for evaluating heart murmur in childhood period, especially when the patient has abnormal facial features or mental problem (Figure 1-5).

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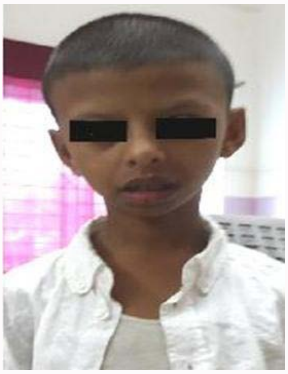

Figure 1: Elfin faces.

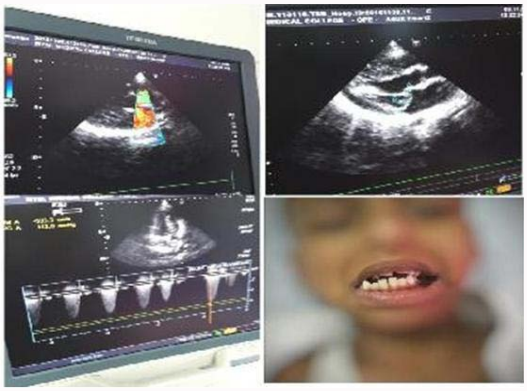

Figure 2: Echocardiogram shows supravalvular aortic Stenosis & MVP.

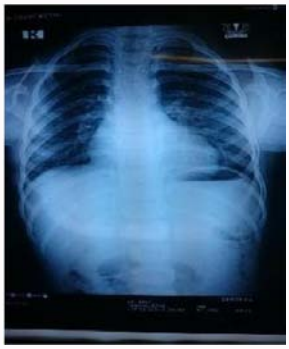

Figure 3: X-ray chest P/A views: Borderline Cardiomegaly.

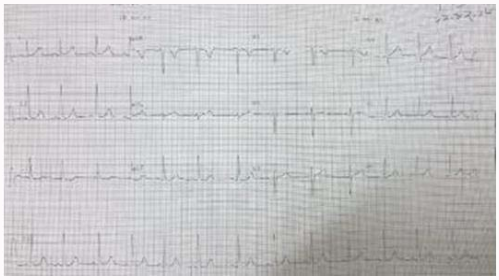

Figure 4: Normal ECG tracing.

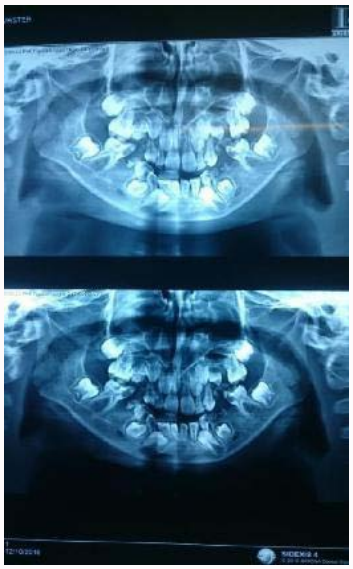

Figure 5: Orthopentogram (OPG) showing multiple malocclude teeth.

Discussion

Williams's syndrome is a rare familial multisystem disorder occurring in 1 per 20,000 live births. It is characterized by congenital heart defects (CHD), Skeletal and renal anomalies, cognitive disorder, social personality disorder and dysmorphic facies. WS is characterized by the triad, supravalvular aortic stenosis (AS), mental retardation and elfin facies. In addition, mild microcephaly, neurologic dysfunction, auditory hyperacusis, narrowing of the peripheral systemic and pulmonary arteries, inguinal hernia, strabismus, epicanthic folds, abnormalities of dental development, high prominent forehead may be present [11]. In addition to typical facial features, patients with WS have a characteristic ebullient personality profile that is classically referred to as the cocktail personality. Patients with WS are hypersocial and garrulous and have a relative verbal strength that belies a mean IQ of 50 to 60 [12]. Patients 50% to 90% meet diagnostic criteria for anxiety disorder, attention deficit-hyperactivity disorder, or phobic disorder [10]. Children with WS are overly social and outgoing, inappropriately friendly to adults and unwary of strangers. Hypercalcemia, which was a prominent features in the report of Bongiovanni et al., [13] has been reported to occur in up to 50% of patients with WS but this usually resolves without intervention in the first two years [14]. WS babies typically have a low birth weight and are often diagnosed as failing to thrive. Cardiovascular defects are the most common cause of death in patients with WS [10]. Structural cardiovascular abnormalities occur in about 80% of all WS patients and are present in up to 93% of WS patients presenting in the first year of life [15]. Although a number of cardiovascular abnormalities are common to WS, the majority consist of some form arterial stenosis. Three anatomical types of supravalvular AS are recognized. The most common is the hourglass type, in which marked thickening and disorganization of the aortic media produce a constricting annular ridge at the superior margin of the sinuses of Valsalva. Uniform hypoplasia of the ascending aorta characterizes the hypoplastic type. Genetic studies suggest that the familial anomaly is transmitted as an autosomal dominant trait with variable expression. The typical appearance is similar to the elfin facies observed in the severe form of idiopathic infantile hypercalcemia [10].

Williams Syndrome is a rare genetic disorder-so rare, in fact, that few people have ever heard of it. Special care needs to be taken when children with WS are given anesthesia. Little babies will come up to you, they will stare into your face and it will be hard to actually disengage from that stare explained Helen Tager-Flusberg whose

lab at Boston University studies the social behavior of children with WS. Genetic diagnosis is confirmed by micro-array analysis & also by the Fluorescent in Situ Hybridization (FISH) test. Multi-specialty approach is needed. WS cannot be cured, but the ensuing symptoms, developmental delays, learning problems & behaviors can be treated. Correction of cardiac defects, cognitive behavior therapy, avoidance of extra Calcium & Vitamin D these are the treatment options.

Conclusion

We present a particular case of Williams Syndrome with low IQ, congenital supravalvular aortic stenosis, stunted growth, eye problems, defective dental enamel and mal-occlusion of multiple teeth. More than 99% of individuals with the clinical diagnosis of WS have this contigous gene delation, detected by FISH.

We should keep in mind and give more attention for evaluating heart murmur in childhood, especially when the patient has abnormal facial dysmorphisom or mental retardation.

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