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Case Report

More than coarctation: Williams syndrome with diffuse arteriopathy

Madamshetty Niharika*, Vaishnavi R. Kendre, Prithi Inamdar, Bhushan Chavan

Department of Pediatrics, D. Y. Patil Medical College, Navi Mumbai, Maharashtra, India

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***Correspondence:**
Dr. Madamshetty Niharika,
E-mail: madamshettyniharika111@gmail.com

ABSTRACT

Williams syndrome is a rare multisystem genetic disorder caused by a microdeletion at chromosome 7q11.23 involving the elastin (ELN) gene. It commonly presents with cardiovascular abnormalities, developmental delay, connective tissue defects, and characteristic facial dysmorphism. We report a 9-month-old male infant with recurrent respiratory tract infections, developmental delay, and failure to thrive. Bronchoscopy showed mild tracheomalacia and bilateral bronchomalacia, and he had previously undergone right inguinal hernioplasty. On examination, characteristic dysmorphic facial features raised suspicion for Williams syndrome. Echocardiography revealed severe discrete coarctation of the aorta with concentric left ventricular hypertrophy and bilateral superior vena cava. During balloon coarctoplasty, right renal artery stenosis was identified, representing a rare vascular association. Genetic studies confirmed the diagnosis of Williams syndrome with deletion involving chromosome 7q11.23. This case highlights the importance of careful clinical evaluation and genetic confirmation in children with syndromic facies and vascular anomalies for early diagnosis and appropriate multidisciplinary management of Williams syndrome.

Keywords: Williams syndrome, Coarctation of aorta, Tracheomalacia, Bronchomalacia, Right renal artery stenosis, Developmental delay, Elfin facies

INTRODUCTION

Williams syndrome is a rare autosomal dominant multisystem genetic disorder caused by a microdeletion involving the elastin (ELN) gene on chromosome 7q11.23.[1] It is characterized by distinctive facial dysmorphism, developmental delay, connective tissue abnormalities, and cardiovascular involvement, most commonly supravalvular aortic stenosis.[2] Elastin arteriopathy in Williams syndrome may affect multiple medium and large vessels, resulting in vascular abnormalities involving the aorta, renal arteries, and other systemic vessels.[2] Most cases arise due to de novo mutations, with an estimated prevalence of approximately 1 in 7,500 to 1 in 20,000 live births.[3] Although cardiovascular manifestations are well recognized, associated airway abnormalities such as tracheomalacia and bronchomalacia are uncommon.[2] Airway involvement may contribute to recurrent respiratory illness. Genetic studies, including exome sequencing, play an important role in confirming the diagnosis. Here, we report a 9-month-old male child with Williams syndrome confirmed on exome sequencing, presenting with developmental delay, recurrent respiratory illness, severe coarctation of the aorta requiring balloon coarctoplasty, and right renal artery stenosis.

CASE REPORT

A 9-month-old male child presented with progressively increasing swelling over the inguinal region since birth, which became more prominent during crying, coughing, and straining. The child had delayed attainment of developmental milestones, feeding difficulties with swallowing incoordination, poor weight gain, and recurrent episodes of cough, noisy breathing, respiratory distress, and lower respiratory tract infections requiring multiple hospital admissions.

At 3 months of age, he underwent right-sided inguinal hernioplasty for progressively increasing inguinal swelling. However, recurrent respiratory symptoms persisted postoperatively. In view of recurrent wheezing, repeated oxygen requirement, and recurrent respiratory exacerbations, flexible bronchoscopy was performed, which revealed mild tracheomalacia and bilateral bronchomalacia.

On examination, the child had failure to thrive and global developmental delay. Dysmorphic facial features suggestive of an underlying syndromic disorder were noted, including broad forehead, bitemporal narrowing, periorbital fullness, depressed nasal bridge, short upturned nose, long philtrum, full cheeks, prominent lips, small chin, and characteristic elfin facies. Peripheral pulse examination revealed feeble lower limb pulses with radiofemoral delay. Blood pressure in the upper limbs was elevated for age compared to the lower limbs.

In view of the dysmorphic facies, developmental delay, recurrent respiratory morbidity, and suspected multisystem involvement, detailed cardiovascular evaluation was undertaken. Two-dimensional echocardiography revealed severe discrete coarctation of the aorta with continuous flow across the descending aorta and a peak gradient of approximately 25–30 mmHg. Concentric left ventricular hypertrophy and bilateral superior vena cava were also noted. The child subsequently underwent successful balloon coarctoplasty.

During angiographic evaluation at the time of coarctoplasty, incidental narrowing of the right renal artery suggestive of right renal artery stenosis was identified, indicating diffuse vascular involvement consistent with elastin arteriopathy.

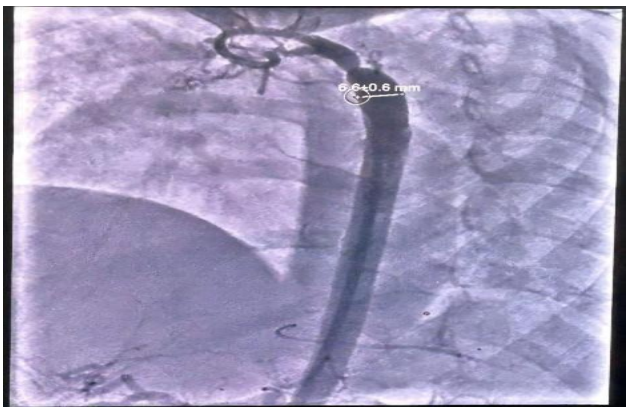

Figure 1: Coarctation of aorta.

The coexistence of characteristic facial dysmorphism, developmental delay, recurrent respiratory illness, airway abnormalities, and cardiovascular involvement raised strong suspicion of an underlying genetic syndrome. Exome sequencing identified a pathogenic microdeletion involving chromosome 7q11.23 with involvement of the elastin (ELN) gene, confirming the diagnosis of Williams syndrome.

This case highlights the importance of careful clinical examination and recognition of syndromic clues in children presenting with recurrent respiratory morbidity, developmental delay, and multisystem involvement. Early diagnosis of Williams syndrome facilitates timely cardiovascular intervention, respiratory management, developmental rehabilitation, genetic counselling, and multidisciplinary follow-up.

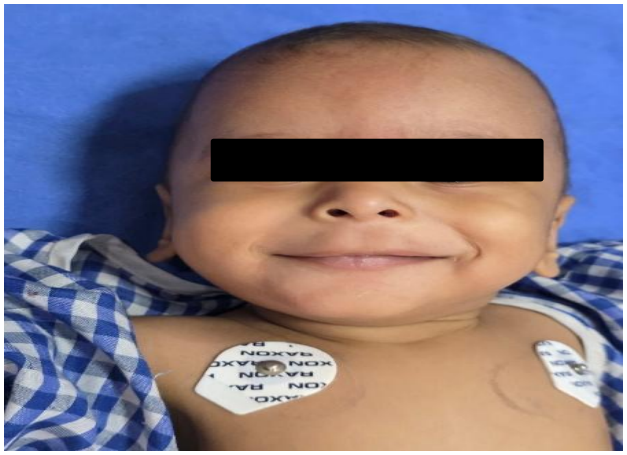

Figure 2: Elfin facies.

Figure 3: Right sided renal artery stenosis.

DISCUSSION

Williams syndrome results from a small deletion on chromosome 7q11.23 that removes the most important gene, the elastin gene.[1,2] Since elastin is a key structural protein in blood vessel walls, airways, and connective tissue, its deficiency leads to the wide range of problems seen in this condition.[1] The targeted genetic testing such as chromosomal microarray or exome sequencing is required to confirm the diagnosis. In our patient, exome sequencing identified the pathogenic deletion at 7q11.23. This confirms the importance of molecular testing in any child with a combination of developmental delay, dysmorphic features, and multisystem disease.

One of the most recognisable features of Williams syndrome is a distinctive facial appearance, often called 'elfin facies'. These features are present in nearly all affected children and are often the first clinical clue that leads a clinician towards the correct diagnosis.[4] In our patient, it was the recognition of this characteristic facial pattern that prompted further genetic evaluation. Children with Williams syndrome also tend to have weak connective tissue, again because of reduced elastin. This shows up as inguinal or umbilical hernias, loose joints, and soft lax skin.[4] The right inguinal hernia in our infant, which required surgery at 3 months of age, fits well within this described pattern of connective tissue involvement and has been reported in other infants with Williams syndrome.

Almost all children with Williams syndrome have some degree of developmental delay, and failure to thrive in infancy is a very common early presenting feature.[5,6] Low muscle tone makes it difficult for infants to coordinate sucking and swallowing, leading to feeding problems and poor weight gain. As they grow older, most children develop mild to moderate intellectual disability, though they tend to have relatively better language and social skills compared to their visuospatial abilities.[5] In our patient, global developmental delay, swallowing incoordination, and failure to thrive were all present from early infancy.

Heart and blood vessel problems are the most serious aspect of Williams syndrome and occur in up to 80% of affected patients.[7] The cause is elastin arteriopathy because elastin in the vessel wall is deficient. The most well-known cardiac lesion is supravalvular aortic stenosis, a narrowing just above the aortic valve, which occurs in more than half of all patients.[7,8] Our patient presented with severe coarctation of the aorta but without evidence of supravalvular aortic stenosis, which is the more classical lesion. This difference from the expected presentation illustrates how much the cardiovascular phenotype can vary between individuals with the same genetic deletion, and why a complete cardiovascular evaluation not just assessment for is essential in every child diagnosed with Williams syndrome. Coarctation of the aorta is a less common but well described vascular manifestation in Williams syndrome. In a study of 41 children with Williams syndrome who had high blood pressure, echocardiography revealed coarctation of the aorta in approximately 9% of patients.[9] Bilateral superior vena cava was an additional finding on echocardiography in our patient. The same arteriopathic process that narrows the aorta in Williams syndrome can also affect the renal arteries, causing renal artery stenosis and renovascular hypertension.[10] In our patient, right renal artery stenosis was not suspected before the procedure but was identified incidentally on aortography performed during balloon coarctoplasty. This finding reinforces the recommendation that when a child with Williams syndrome undergoes cardiac catheterisation, a complete aortogram including visualisation of the renal arteries should always be performed. Airway problems such as tracheomalacia and bronchomalacia are infrequently reported in Williams syndrome and are likely under-recognised in clinical practice. Because elastin is also a structural component of the airway wall, its deficiency can lead to abnormal softness of the trachea and bronchi, causing them to collapse during expiration. In our patient, flexible bronchoscopy confirmed mild tracheomalacia with bilateral bronchomalacia. These airway findings explained the persistent respiratory symptoms, repeated hospital admissions, and ongoing oxygen requirement that had continued despite adequate cardiac and general medical management. This case supports routine consideration of flexible bronchoscopy in any infant with Williams syndrome who has unexplained or treatment-resistant respiratory symptoms, rather than assuming all airway symptoms are secondary to cardiac disease.

The combination of severe coarctation of the aorta, right renal artery stenosis, tracheobronchomalacia, and bilateral superior vena cava occurring together in a single infant with genetically confirmed Williams syndrome has not been previously reported in the literature. This rare combination makes this case clinically noteworthy, as it demonstrates the breadth of multisystem involvement that elastin deficiency can produce and highlights the need for thorough investigation across cardiac, vascular, airway, and renal systems in every child with this diagnosis.

CONCLUSION

This case highlights the importance of careful clinical examination and early recognition of syndromic clues in children presenting with recurrent respiratory illness, developmental delay, and cardiovascular abnormalities. The coexistence of severe coarctation of the aorta, renal artery stenosis, and airway abnormalities in Williams syndrome emphasizes the variable multisystem involvement seen in this condition. Early diagnosis with genetic confirmation allows timely intervention and appropriate multidisciplinary management.

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