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Case Report

Williams syndrome in a 7-year-old girl with characteristic facies, congenital cardiac involvement and behavioral phenotype: a case report

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ABSTRACT

Williams syndrome is a rare multisystem genetic disorder caused by a microdeletion at chromosome 7q11.23 involving the ELN gene. It is characterized by distinctive facial features, cardiovascular abnormalities, developmental delay, endocrine disturbances, and a characteristic hypersocial behavioral profile. We report a 7-year-old girl with genetically confirmed Williams syndrome who presented with urinary tract infection. She had been diagnosed at 1 year of age after recognition of dysmorphic features and congenital cardiac disease, followed by confirmation by chromosome analysis and fluorescence in situ hybridization. Clinical findings included depressed nasal bridge, posteriorly rotated ears, long philtrum, retrognathia, mild pectus deformity, deep-set nails, and mild muscular ventricular septal defect. At follow-up, she had typical "elfin" facies and an unusually friendly personality. This case highlights the importance of early recognition and long-term multidisciplinary follow-up, including cardiovascular, developmental, behavioral, metabolic, renal, hearing, and visual surveillance.

Keywords: Williams's syndrome, Williams-Beuren syndrome, Elfin facies, Congenital heart disease, Developmental delay

INTRODUCTION

Williams syndrome, also known as Williams-Beuren syndrome, is a rare contiguous gene deletion disorder caused by a heterozygous microdeletion of chromosome 7q11.23, usually including the ELN gene. It has an estimated prevalence of about 1 in 7,500 to 1 in 20,000 live births and is recognized as a multisystem disorder involving the cardiovascular, neuro-developmental, endocrine, connective tissue, renal, and behavioral domains.[1-3] Characteristic features include distinctive facial appearance, developmental delay, mild to moderate intellectual disability, a unique cognitive profile with visuospatial weakness, cardiovascular disease, and a markedly social personality often accompanied by anxiety and attention difficulties.[4]

Cardiovascular disease is one of the most clinically significant components of Williams syndrome. Elastin arteriopathy may manifest as supravalvar aortic stenosis, peripheral pulmonary artery stenosis, hypertension, and other vascular lesions, and it remains a major source of morbidity across the lifespan.[5] In addition, affected children may show hypercalcemia or hypercalciuria, feeding difficulties, hearing sensitivity, ophthalmic abnormalities, and a broad range of developmental and psychiatric concerns.[6,7] Early clinical suspicion followed by molecular confirmation is therefore important not only for diagnosis but also for planning structured surveillance.

We report a 7-year-old girl with genetically confirmed Williams syndrome whose case demonstrates the classical combination of dysmorphic facies, congenital cardiac involvement, and behavioral phenotype, while also

highlighting the importance of long-term multidisciplinary follow-up.

CASE REPORT

A 7-year-old girl, a known case of Williams syndrome, was brought to the outpatient department by her mother with symptoms suggestive of urinary tract infection. At presentation, she had characteristic "elfin facies" and a very friendly personality. She was already on regular follow-up and underwent annual medical evaluation, blood pressure assessment in both arms, vision and hearing screening, urinalysis, and spot urine calcium-to-creatinine ratio testing. She was also under psychological and psychiatric follow-up for attention-deficit/hyperactivity disorder and anxiety.

Her diagnosis had been established at 1 year of age after clinical suspicion arose because of a constellation of findings, including mild muscular ventricular septal defect, depressed nasal bridge, posteriorly rotated ears, mild pectus deformity, long philtrum, retrognathia, and deep-set nails. Chromosome analysis and fluorescence in situ hybridization confirmed Williams syndrome (Figure 1).

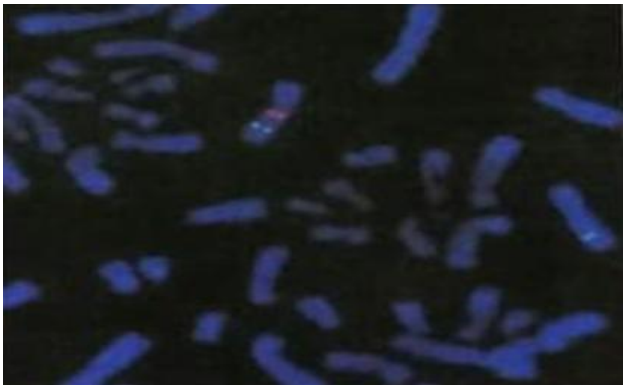

Figure 1: Fluorescence in situ hybridization. FISH was performed on metaphase chromosomes from cultured peripheral blood sample of this patient using LSI William syndrome (elastin gene) region probe localized to 7q11.23. Probe hybridization showed the signal only on one of the chromosome 7 at 7q11.23 region (red) in all the metaphases analysed.

The child's phenotype was consistent with the recognized clinical spectrum of Williams syndrome. The facial dysmorphism, congenital cardiac lesion, and unusually social personality fit well with the syndrome described in standard references and recent reviews.

Although the current visit was prompted by urinary tract infection, the syndrome itself remained central to management. Children with Williams syndrome require ongoing surveillance for cardiovascular abnormalities, hypertension, calcium disturbances, renal complications, developmental delay, and psychiatric comorbidity. The annual use of urinalysis and calcium-related monitoring in this child was therefore clinically appropriate.

No acute cardiovascular compromise, seizure activity, or major metabolic instability was documented in the available summary at this visit. Management was directed toward the intercurrent urinary tract infection while continuing syndrome-specific long-term follow-up.

DISCUSSION

This case illustrates several classical and clinically relevant features of Williams syndrome. First, the diagnosis was suspected in infancy on clinical grounds before molecular confirmation. That pattern is common because the syndrome often becomes recognizable through the combination of characteristic facies, developmental concerns, congenital heart disease, and a striking behavioral profile.[8] Depressed nasal bridge, long philtrum, retrognathia, and later-described elfin facies in this child fit well within the known phenotype.[9]

Second, congenital cardiac involvement remains one of the most important diagnostic and prognostic aspects of Williams syndrome. Although supravalvar aortic stenosis is the classical lesion, a broader cardiovascular spectrum is recognized, and continued surveillance is recommended even when the initial defect is mild.[10] In this child, mild muscular ventricular septal defect contributed to early recognition and justified ongoing follow-up.

Third, the behavioral profile in this case was highly characteristic. The child was described as very friendly and was under treatment for ADHD and anxiety. Recent psychiatric reviews note that Williams syndrome is associated with hypersociability, anxiety, attentional dysregulation, and a distinct social-cognitive phenotype that may significantly affect schooling and daily functioning.[11] Early developmental, psychological, and psychiatric support is therefore essential.

Fourth, the case underscores the importance of multidisciplinary surveillance. Standard follow-up in Williams syndrome includes blood pressure monitoring, cardiovascular assessment, developmental review, and evaluation for calcium disturbances, hearing problems, and visual abnormalities.[6] This child's periodic blood pressure measurement in both arms, urinalysis, calcium monitoring, hearing and vision assessment, and neurobehavioral follow-up were appropriate and reflect good long-term care.

Finally, early molecular confirmation was valuable. Fluorescence in situ hybridization confirmed the diagnosis in infancy in this child, allowing timely counseling and organized surveillance.[12] Although chromosomal microarray is also widely used now, molecular confirmation remains important for diagnostic certainty and anticipatory care.

CONCLUSION

This case highlights the classical multisystem nature of Williams syndrome and the importance of recognizing the disorder from the combination of dysmorphic facies, congenital cardiac findings, developmental and behavioral abnormalities, and a characteristic friendly personality. Early diagnosis allowed structured follow-up in this child, including cardiovascular, renal, metabolic, sensory, and neurobehavioral surveillance. Williams syndrome should be considered in children with compatible facial features and congenital heart disease, especially when accompanied by unusual sociability and developmental concerns.

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