Case Report

Cleft palate in Williams syndrome

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ABSTRACT

Williams–Beuren syndrome (WBS) is a genomic neurodevelopmental disorder, estimated to occur in approximately 1 in 10,000 persons. It is caused by a deletion of the "elastin" gene on chromosome 7q11.23 and was described officially in 1961 by Williams, Barrat-Boyes, and Lowe. Cleft palate is not considered in the medical literature as a part of the multisystem disorders of the Williams syndrome but it was yet described. We present our experience of a patient who presents cleft palate among other congenital malformations.

Keywords: Cleft palate, congenital malformations, Williams–Beuren syndrome

INTRODUCTION

Williams–Beuren syndrome (WBS) was first recognized as a distinct entity in 1961. It is present at birth, and affects males and females equally. It can occur in all ethnic groups and has been identified in countries throughout the world, with an estimated prevalence of 1 in 10,000 births. Williams syndrome is a genomic disorder, caused by the micro-deletion of about 26 genes from the long arm of chromosome 7, in the region that is now known as the "WBS chromosome region". The diagnosis is generally established at birth with the fluorescent *in situ* hybridization (FISH) test. WBS is characterized by multisystem disorders: Cardiovascular anomalies, such as supravalvular aortic stenosis (SVAS) and hypertension, a distinctive "elfin" facial appearance with mental disability and a very characteristic highly social behavior.[1] Cleft palate has never been considered part of the WBS but the condition has not been yet described in medical literature.

CASE REPORT

The patient was presented to us, for the first time, at the age of 1 year. He is the only son of a family with no previous history of congenital malformations. At birth he presented characteristic clinical features like the bitemporal narrowing, the epicanthal fold, and the flat nasal dorsum [Figure 1]. Moreover, this patient presented a SVAS and renal abnormalities and rare a U-shape cleft palate. Hence diagnose of Williams syndrome, based on the clinical features and supported by the FISH test was established. At the age of 9 months he underwent surgery for the correction of an inguinal hernia. At the age of 13 months we approached a palatoplasty through the Kriens technique and he was discharged on the first postoperative day in a good loco-regional condition.[2] The patient was treated during the humanitarian mission "Sorrisi in Italia", organized by the charity foundation Operation Smile.

DISCUSSION

Initial appreciation of the syndrome came from two seemingly unrelated disorders: Idiopatic hypercalcemia of infants, largely the domain of pediatricians, and SVAS, treated by cardiologists. Then Anna Blancquaert, Belgian pediatric cardiologist, was the first to point out the similarity of the facies in patients with idiopathic hypercalcemia of infancy and those with SVAS and mental retardation.[3] In 1961, J.C.P. Williams described for the first time a new unrecognized syndrome, the association of SVAS and mental retardation with distinctive facial appearance and "friendly" personality in four nonconsanguineous children.[4] A year after this report, German physician Dr. A. J. Beuren described three new patients with the same presentation.

Later WBS was accurately described and nowadays it is commonly known that it is caused by a microdeletion at 7q11.23., now

referred as WBS chromosome region, spanning 1.5-1.8 million base pairs and containing 26-28 genes.[5]

FISH involving Elastin gene (ELN)-specific probes, establishes the diagnosis for this syndrome by showing the presence of a single ELN allele only rather than two alleles. WBS is estimated to occur in approximately 1 in 10,000 persons. WBS has a characteristic constellation of clinical features that include: unusual facies, abnormal behavioral abilities, cardiovascular anomalies, renal, and other abnormalities. Cleft palate has never been referred to be part of this multidisorders syndrome; we do not know the frequency of Williams' syndrome association with cleft lip or palate because of the lack of the medical literature.[6] We approached the cleft palate repair through the Kriens palatoplasty[2] [Figure 2]. This technique consists of a three layer palatoplasty with intravelar veloplasty: Palatal muscle reconstruction offers better velopharyngeal competence and eustachian tube function. The levator muscle repositioning procedure or intravelar veloplasty during palatoplasty is the most widely practiced method to achieve velopharyngeal competence. Victor Veau first described the 'cleft muscles' and advocated the concept of midline levator palatini muscle reapproximation. He emphasized the importance of an encircling suture to pull the levator muscle bundles together, side to side. Braithwaite and Kriens further improved this technique. They emphasized careful dissection of abnormally positioned levator muscles and the need to free the levator palatini from the posterior edge of the hard palate to restore the levator sling and allow tension free closure in the midline. The operation is technically challenging, and there is great variability in the degree of muscle dissection and overlap across the midline.[7] Hence, results vary among surgeons. The patient was discharged on the first postoperative day and he did not show any major/minor complications [Figure 3].

The patient's facial features are typical. He presents bitemporal narrowing, epicanthal fold, prominent ears, enlarged philtrum, flat nasal dorsum, malar hypoplasia, and puffiness around the eyes. Moreover, these patients have characteristic attitude of the mouth, which is maintained in the open position, stellate/lacy iris pattern, strabismus, and full cheeks.[8] It results in characteristic "elfin-like" face. Cardiovascular anomalies are usually represented by SVAS with arterial narrowing: The aortic root is dilated and the proximal ascending aorta is mildly hypoplastic. This patient presents a SVAS. SVAS is rare outside the setting of WBS, except in familial SVAS syndrome. It occurs in approximately 70% of the patients affected by this syndrome. The arterial narrowing is due to thickening of the vascular media from smooth muscle overgrowth and can lead to stenosis of medium and large arteries. This arterial condition is also known to occur in numerous locations, including aortic arch, the descending aorta and the pulmonary, coronary, renal, mesenteric and intracranial arteries. An increased carotid artery intima-media thickness consistent with generalized elastin arteriopathy is present in all cases. Hypertension usually develops in about 50% of patients. Cardiovascular complications are the major cause of death in these patients: There is a cardiovascular-associated mortality 25-100 times than among the controls.[9] Other important clinical features are represented by the behavioral aspect; this patient showed a very friendly behavior. This is characteristic of Williams' syndrome patients; they are described as highly sociable, approachable, overly friendly, never going unnoticed in a group, and empathic. This combination of characteristics, which has not been attributed to any other syndrome or to children who are developing normally, is usually defined as "cocktail-party syndrome".

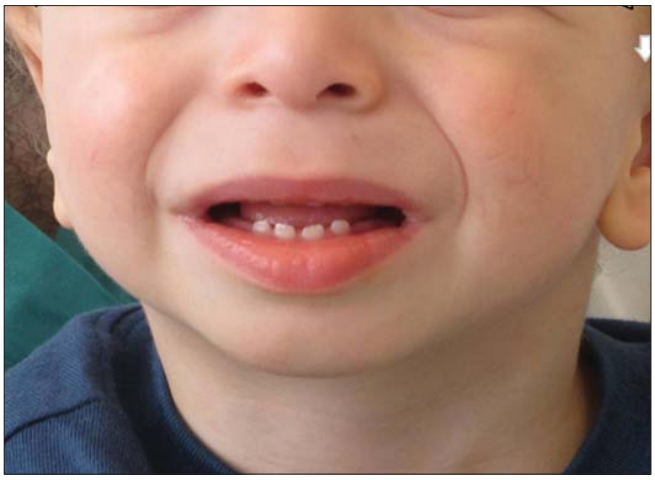

Figure 1: Preoperative frontal view

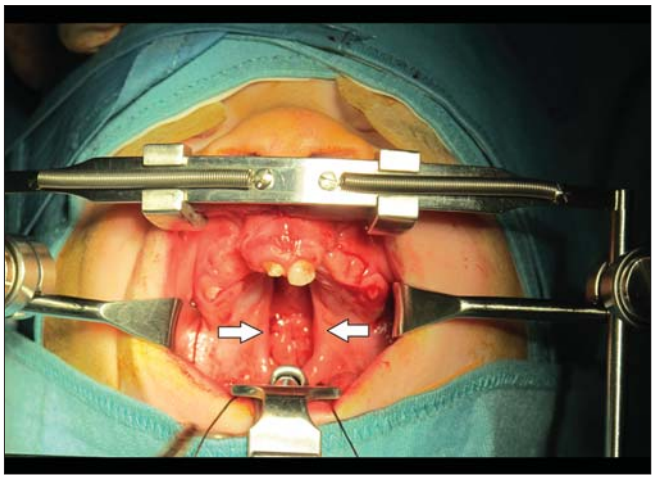

Figure 2: Intraoral preoperative view

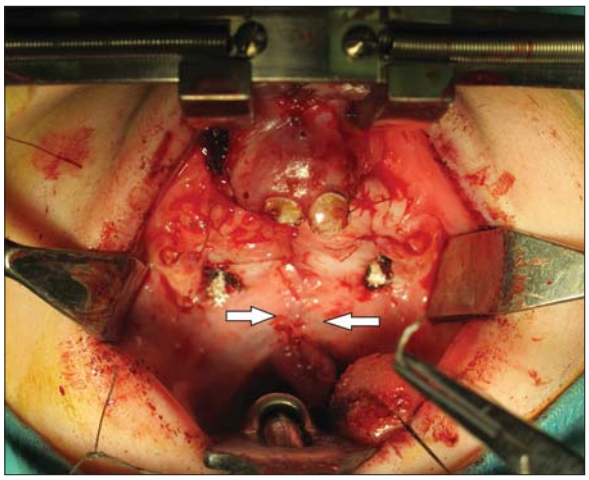

Figure 3: Intraoral postoperative view

As seen in the literature, the patient presents low muscle tone and joint laxity. As the children get older, joint stiffness (contractures) may develop.

Despite the frequent association with hypercalcemia, our patient reveals normal calcium serum level.

Williams syndrome is associated with an abnormal pattern of growth. In the first 4 years of life, weight gain and linear growth are poor. Our patient shows, in fact, a height that is below the third percentile. Even though there is improvement in growth in mid-childhood, 70% remain below the third percentile for mid parental height and are thus shorter than would be expected for their genetic background. This combination of factors leads to a mean final adult height that is below the third percentile.[10]

CONCLUSION

The primary care physician remains the principal provider and care coordinator for patients with WBS. Treatment involves a combination of medical monitoring, anticipatory guidance, direct therapies, pharmacotherapy, surgery, and adaptive changes. None of the available treatments are curative. Cleft palate has been reported in literature just in a few cases as part of the multidisorders features that characterized this syndrome.[11] Thus our patient represents an important rarity.

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