Atrial myxoma and Williams-Beuren syndrome. An incidental association?

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Abstract

We report the case of a 15 years old girl with Williams-Beuren syndrome and atrial mixoma.

Background

Williams-Beuren syndrome (WBS) is caused by genomic microdeletion at human chromosome 7q11.23 (Mouse 5G2), resulting in various cardiovascular, developmental, metabolic, and mental disorders.[1] This microdeletion, which generally occurs sporadically, results in the loss of many genes, including the elastin gene. Vascular malformations require regular follow-up as well as dedicated management. For this reason, children affected by WBS should be regularly followed by paediatric cardiologist with specify expertise in rare disease.[2] The management of arterial hypertension requires a combination of pharmaceutical treatment with a healthy diet and lifestyle. The decision to opt for surgical repair of renal artery stenosis must take into account the global involvement of the vascular walls in this pathology. Educational management of children affected by Williams syndrome should be proposed in a multidisplinary setting involving paediatricians, orthodontists, psychomotricists, speech therapists and psychologists.[2] WBS individuals have a combined prevalence of cardiovascular disease of 84% that includes supra- and sub-aortic stenosis (SVAS); aortic, pulmonary, and mitral valvular disease; aortic coarctation; hypertension; and, less commonly, myocardial infarction.[3] To date, cardiac tumors have not been reported in patients with WBS.

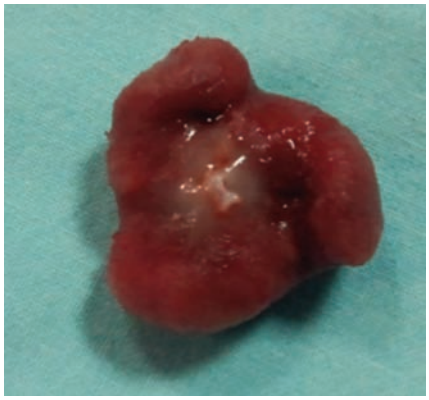

Figure 1. Gross appearance of the myxoma which was removed during surgery.

Case Report

We report the case of a 15 years old girl who had a fetal diagnosis of mitral dysplasia. At birth, a mitral dysplasia with aortic coarctation was detected by ultrasonography and she underwent surgical correction (decoarctation with patch). At 2 yo, for facial dysmorphysms, failure to thrive, and cognitive development, a clinical diagnosis of WBS was suspected, and then confirmed by FISH analysis. At 7 years, a balloon angioplasty was performed for aortic re-coarctation. At 10 years, following the evidence of severe mitral regurgitation, she underwent mitral surgical valvuloplasty with residual moderate regurgitation. At 13 yo, a posterior mitral leaflet prolapse with severe regurgitation was detected by echocardiography, requiring a new annuloplasty with rigid prosthetic rings. After the last operation she was followed up by semestral clinical and echo evaluations, showing trivial-moderate mitral regurgitation, dilated left atrium, normal isthmus flow. At last follow-up, a 3x2 cm mass was identified on the right atrium, attached to the edge of tricuspid valve. The mass was prolapsing through the tricuspid valve into the right ventricle without obstructing the inflow. The mass was excised, and histological evaluation revealed an atrial myxoma (Figure 1).

Discussion

To our knowledge, this is the first report of an atrial myxoma in patients with WBS. Cardiac abnormalities in WBS include supra- and sub-aortic stenosis (SVAS); aortic, pulmonary, and mitral valvular disease; aortic coarctation; hypertension; and, less commonly, myocardial infarction. Cardiac tumors are rare cardiac abnormalities with an incidence of less than 0.1 percent, and they have previously not been reported associated to WBS. On the other hand, patients with other syndromes (such as RAS-MAPK syndromes: Costello and cardiofaciocutaneous syndrome) are at increased risk of cardiac masses. It is not known whether atrial mixoma is a bystander in WBS, or may be part of the clinical spectrum of the disease. WBS results from deletions involving the elastin gene on chromosome 7q11.23. This gene's protein product gives blood vessels the stretchiness and strength required to withstand a lifetime of use. The elastin protein is produced only during embryonic development, infancy and childhood, when blood vessels are formed. Because they lack the elastin protein, people with WBS have disorders of the circulatory system and heart defects. It is not clear if a deficit of elastin may favor cardiac mass development in these patients.

Conclusions

We described the case of a 15 y.o. girl with WBS and atrial mixoma. This is the first case reporting this clinical association, and the association is likely to be a random rather than part of the WBS spectrum.

References

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