GENETICS AND GENODERMATOSES

EHLERS-DANLOS SYNDROME WITH INVOLVEMENT OF HEART VALVES

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Background: Ehlers-Danlos syndrome (EDS) comprises a group of hereditary connective tissue disorders whose main clinical features are hyperextensibility of skin, hypermobility of joints and generalized fragility of connective tissue. It can be classified into classic, hypermobile, vascular, kyphoscoliotic, arthrochalasia and dermatosparaxis. Cardiac valve involvement is more commonly observed in the cardiac valvular subtype, a rare variant of phenotypic overlaps with other EDS subtypes, associated with mutations in type I collagen. Conditions with severe cardiac valve involvement are predicted in only 6% of classic cases of mild to moderate form.

Observation: Female patient, 10 years old, diagnosed with EDS for 9 years, based on clinical criteria and complementary tests. At 1 year of age, she had delay of neuropsychomotor development, bilateral lower eyelid edema, bluish sclera, hyperextensible skin, hypermotility joints and bilateral umbilical and inguinal hernias. Karyotype presented genotype 46 XX, and echocardiogram without alterations. At the moment she presents with articular hypermotility, smooth skin, velvety, hyperextensible, aged face and systolic murmur 1 + / 4 +. The echocardiogram changed showing important alterations: mitral valve prolapse, major mitral insufficiency, mild tricuspid insufficiency and minimal aortic insufficiency. The electrocardiogram showed alterations compatible with right ventricular overload. The patient was also diagnosed with hiatal hernia and Zenker diverticulum. The child has not undergone molecular DNA analysis because such a diagnostic feature is not available at our service.

Key message: Characteristics reported by the patient are compatible with EDS. However, this case has become even more valuable for its exuberance and severe impairment of heart valves, which is rare. Therefore, greater accessibility of the population to the exams is essential, especially the genetic tests as well as multidisciplinary monitoring to prevent possible complications.