Ehlers Danlos syndrome: A case report

Abstract

Ehlers-Danlos syndrome (EDS) is a group of hereditary collagen diseases characterized by joint hyperlaxity, skin hyperelasticity, and generalized tissue fragility. We present the case of an 8-year-old child with EDS in its arthrocalasic form type VII, according to the Villefranche classification, who was born to first-cousin parents. There is no curative treatment for EDS, but it is important to make an early diagnosis for optimal symptomatic management of patients and prevention of avoidable complications.

Keywords: Ehlers-Danlos syndrome,arthrocalasic(arthrochalasia)form,Villefranche classification

Introduction

EDS is a heterogeneous group of inherited connective tissue diseases that present clinically as skin hyperelasticity, joint hypermobility, atrophic scarring and blood vessel fragility (1) (2).

It affects all organ systems, which can lead to significant morbidity and mortality.

An updated classification with 13 types of Sed (EDS)was published in 2o17 and is now the gold standard (2).

It is important to identify the type of EDS to guide management and counseling (1).

The purpose of this article is to describe the arterioachalic type and to show the importance of a collaborative approach to the care of patients with this syndrome.

Case Presentation

An 8 year old child, of 1st degree consanguineous parents, who presents since birth a hypotonia, a thin and pasty skin giving an aspect of cardboard skin and a congenital dislocation of the bilateral hip rebelling to orthopedic treatments with a delay of motor acquisition.

On clinical examination, the child had a delay in growth and weight, with a height of 110 cm (-2DS), a weight of 18 kg (-2DS), and a cranial perimeter of 52 cm. He had facial dysmorphia, a globular thorax, a very severe amyotrophy showing his skeleton (figure 1), and ligament hyperlaxity (figure 2) with skin hyperextensibility (figure 3). Sitting was possible; the child could not get up from a lying position without help; standing and walking were impossible. He had severe hypotonia. Osteotendinous reflexes were present but much attenuated. Babinski was indifferent.

Thyroid hormone levels, muscle enzymes, brain imaging, and echocardiography were normal.

The electroneuromyogram showed normal motor and sensory nerve conduction in the nerves explored. Except for a rich tracing in relation to the effort at the level of the right/left biceps brachii, elsewhere normal tracing in relation to the effort.

Odontologically, there was ameloid hypoplasia at levels 11 and 21 with severe polycaria. Radiographic examination showed taurodentism in the malar 4 teeth of a 6 year old. He had no periodontitis.

According to the Villefranche classification, the most probable diagnosis is EHLERS-Danlos syndrome in its arthrocalasic form type VII.The child had benefited from a genetic consultation. The genetic study of this heterogeneous syndrome is not yet available in Morocco.

DISCUSSION :

The prevalence of Ehlers-Danlos syndrome is estimated to be between 1 in 5,000 and 1 in 100,000, depending on the DHS subtype, but this may be an underestimate (3). The precise prevalence of the different subtypes of EDS is still not known; it is estimated that the achalasia form is extremely rare.

EDS is a heterogeneous family of genetic connective tissue disorders that share the following clinical triad:

-Skin hyperelasticity that is objectified by pinching and pulling of the skin that returns to its original shape after relaxation. (4)

It is the most common form of hip disease in the world, with repeated dislocations and subluxations, complicated by chronic pain and early osteoarthritis. It manifests itself by dislocations of the hip in newborns. The evaluation of this joint hyperlaxity is carried out using the Beighton scale, where a score greater than or equal to 5 out of 9 indicates joint hypermobility (5).

-Tissue fragility with the appearance of a hematoma in the event of a benign trauma with atrophic scars and thin skin.Mental development is usually normal.

These signs are positive to varying degrees in each type of EDS. Patients with EDS may have valve prolapse by pillar rupture that should be routinely sought.

Patients may have craniofacial abnormalities, and in the study by Hagberg et al, periodontal involvement is present in 34 of the SEDs, such as hypodontia of permanent teeth, delayed eruption, and dentinal dysplasia (6). A lack of attached gingiva may be a pathognomonic feature (7). Thus, dentists have a crucial role in early diagnosis and management.
(8)Diagnosis relies heavily on clinical symptoms, so imaging such as echocardiography and MRI is useful in assessing common cardiovascular problems such as mitral valve prolapse and aortic dilatation.
The pathophysiology of most subtypes of Ehlers-Danlos syndrome involves inherited mutations in collagen synthesis and/or processing. Arthrochalastic EDS is caused by heterozygous mutations in col 1A1 or col 1A2 that cause total or partial loss of the cell exon of the respective gene, hence the use of genetic study for subtype-specific diagnosis.

Some diseases, such as Marfan syndrome, fibromyalgia, osteogenesisimperfecta, chronic fatigue syndrome, or depression, may pose a problem of differential diagnosis with EDS.There is no curative treatment, so the management is global and symptomatic, based on care, avoidance of violent exercises, and physical therapy sessions in order to prevent complications (vascular and organic rupture, hemorrhagic risk, sleep disorders, and sleep breathing disorders) (4).

Conclusion

EDS is a model of pathology whose classification is still evolving, so that EDS is no longer solely associated with collagen mutations but also with other constituents of the extracellular matrix.These studies allow us to identify new therapeutic targets.

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Figure 1: Chest deformity and amyotrophy

Figure 2:Dorsiflexion of the finger over 90

Figure 3:Skin hyperextensibility: