

Cutis laxa - Case report *

Cútis laxa - Relato de caso

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Abstract: Cutis laxa is a rare inherited or acquired disorder of elastic tissue characterized by inelastic and loose skin. Congenital cutis laxa may present with internal organ involvement, determining a worse prognosis. The authors present the case of a female patient with clinical manifestations suggestive of the hereditary form of the disease, with consanguineous parents (second-degree cousins) and a brother who died with a similar clinical presentation. The genetic study of the FBLN5 gene was important to confirm the diagnosis, define the prognosis, and provide genetic counseling to the family.

Keywords: Consanguinity; Cutis laxa; Genetic counseling

Resumo: A Cútis laxa é uma doença rara, hereditária ou adquirida. Resulta da alteração do tecido elástico, tornando a pele frouxa e inelástica. A forma congênita tem geralmente acometimento sistêmico, com pior prognóstico. Os autores relatam o caso de uma paciente, com quadro clínico sugestivo da forma hereditária da doença, com pais consangüíneos (primos de 2º grau) e irmão falecido com clínica semelhante. O estudo genético do gene FBLN5 teve importância na confirmação diagnóstica, na definição do prognóstico e no aconselhamento genético familiar.

Palavras-chave: Aconselhamento genético; Consangüinidade; Cútis laxa

INTRODUCTION

Cutis laxa is a rare disorder of connective tissue. It is caused by an abnormality in elastic fibers, which are essential to the structural integrity of various tissues, including the skin. ¹

CASE REPORT

Female patient, white, 33 months old, was admitted to the hospital with symptoms suggestive of bronchopneumonia. When the patient was 1-year-old, she presented dyspnea and was diagnosed with pulmonary emphysema, making the continuous use of oxygen at home necessary. Her parents first noticed that she had looser skin when she was 7 months old. She had a history of several hospital stays due to bronchiolitis, respiratory insufficiency, spontaneous pneumothorax, and cystitis. She had been previously

submitted to four right and one left inguinal herniorrhaphy procedures. She had a normal psychomotor development; ponderal index in the 10 percentile and length index below the 5 percentile; consanguineous parents (second-degree cousins) and a brother who died at nine months with a similar clinical presentation. Physical examination revealed a dyspneic, cyanotic, and hoarse patient. The skin of her face was pendular with sagging folds; she showed left eye ectropion and right eye entropion (Figures 1 and 2), associated with conjunctival hyperemia and tearing. Her nose was flat and wide and the skin of her body was flaccid. Pulmonary exam revealed diffuse snores and murmurs, subcostal retractions. A genetic study of the family was conducted and a homozygous mutation in the FBLN5 gene was found, responsible for the autosomal recessive form of cutis laxa.

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FIGURE 1: Loose skin of the face, with sagging folds and aged aspect



FIGURE 2: Lateral view of the face

DISCUSSION

Cutis laxa is a rare disorder that occurs due to alterations in the elastic tissue caused by defects in its synthesis or by its destruction. It may be hereditary or acquired.^{1,2} Its typical dermatologic characteristics are loose skin with folds and premature aged appearance. Loss of skin elasticity may involve the entire skin surface, but it is more evident on the face, neck, dorsum and thighs.¹ Systemic manifestations may be associated, including large vessel aneurysms, pulmonary emphysema, pulmonary artery stenosis, hernia, and diverticula in the gastrointestinal and genitourinary tracts, which determines a worse prognosis. Involvement of internal organs is more frequent in the autosomal recessive hereditary form of the disease, in which skin manifestations are often present at birth.³ The patient showed, in addition to these skin manifestations, pulmonary emphysema and history of inguinal hernias that needed several surgical corrections.

Over the last decade, genetic research has advanced to help us understand the etiology of the hereditary form of cutis laxa. Different patterns of transmission have been identified, such as the autosomal dominant form (mutations in the elastin gene),

the X-linked recessive variant (ATP7A gene, copper transporting ATPase), and type I autosomal recessive form (mutation of the FBLN5 gene, codifier of fibulin-5, a protein associated with the elastic fiber).²

Cutis laxa is diagnosed by clinical and histological (skin biopsy) examination. The latter shows reduced, irregular and fragmented elastic fibers in the reticular layer of the dermis.⁴ Currently, with new discoveries, genetics has been used to confirm the diagnosis.^{2,5}

This case was diagnosed through clinical examination and it was molecularly confirmed by the identification of the c.850C>T mutation (exon 8 of the FBLN5 gene), causing the aminoacid that would normally be codified in the 284 position of the protein (arginine) to be replaced by stop codons. This generates an abnormality in the formation of fibulin-5 and, consequently, in the elastic fiber. This mutation, observed in homozygous form (which conforms to the fact that the parents are consanguineous), has never been described before.

Information about the genotype is important for a conclusive diagnosis, better understanding of the prognosis, and genetic counseling of the family. □

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