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Cutis Laxa syndrome a case report

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Abstract

Introduction

Cutis laxa syndrome is a group of heterogeneous affections of elastic and connective tissue, Etiopathogenesisis poorly known, It may be congenital or acquired, it is very rare, responsible for a skin hyper laxity associated with multi-organ involvement which defines the prognosis.

Case report: We describe the observation of a newborn in post-term, at H1 of life; from a non-consanguineous marriage, Coming from a poorly monitored pregnancy; 20-year-old mother, there is no taking medication during the pregnancy; with positive infectious history. The delivery was vaginal, Apgar 10/10. He was admitted to the intensive care and neonatology department for immediate respiratory distress3/10 according to Silverman score. With the clinical examination find a poly-malformative syndrome associating a stretchy skin that stretches evoking the cutis laxa, and ligament hyper mobility, with a hoarse voice associated with the multi-organ involvement of inguino-scrotal hernia and a major hiatus hernia.

Discussion

Cutis laxa syndrome brings together a group of extremely rare conditions; it's a condition that can be acquired or congenital. There are three clinical forms depending on the mode of transmission: The autosomal dominant forms, The autosomal recessive and the X-linked form, forms acquired in neonatology may be due to taking medication during pregnancy, or due to fever, inflammation or autoimmune disease. In our case there is no notion of the mother taking medication, our patient's clinic corresponds more to the autosomal recessive form, given the cutis laxa associated with multi-organ involvement, inguino-scrotal hernia and ligament hyper mobility and hoarse voice. Hiatal hernia is often associated with this form. The mutation of the gene responsible for this form is identified as fibulin 5gene, our genetic study is ongoing.

Keywords: cutis laxa - Hiatal hernia - Connective tissue.

Introduction

Cutis laxa syndrome is a group of heterogeneous affections of the elastic and connective tissue, characterized by skin hyper laxity. In Latin CUTIS LAXA means LOOSE SKIN. It is associated with variable systemic manifestations. It can be congenital or acquired; it's a very rare condition. Only a few rare cases have been reported in the literature. However, it can be estimated that there are probably less than 1000 cases in the world; we describe a new observation of a newborn baby at h1 of life.

Case report

It is a newborn child in post-term, from a non-consanguineous marriage and whose parents are in good health, there are no similar cases in the family, issuing from a poorly monitored pregnancy; 20-year-old mother, no history, no notion of taking medication during pregnancy; G1P1; positive infectious history, the delivery was vaginal; Apgar 10/10 weigh of birth 4kg600, height to 50 cm, head circumference at 36 centimeters. He was admitted to the intensive care and neonatology department for immediate respiratory distress at 3/10 according to Silverman score.

The clinical examination at birth found a facies particularly suggestive of cutis laxa, with a stretchy skin that stretches easily and numerous flabby folds in the face, the newborn was cyanotic saturated at 70%with ambient air, hypotonic, poorly reactive, with a weak sucking reflex, a GSC 15/15, temperature at 37°, heart rate at 150 beats per minute and respiratory rate at 60 cycle/min. The pleuropulmonary examination found a thorax of normal morphology, intrathoracic hydroaeric noise, with a Silverman score of 3/10.The cardiovascular examination was normal. As well as a poly-malformative syndrome involving a hoarse voice, ligament hyper mobility, an inguinoscrotal hernia and a hiatus hernia on the chest X-ray on admission. Biologically: KTVO was negative, H24 was positive, CRPat 57g/, with negative Lumbar Puncture, without any other disorder, the patient was treated as an early bacterial neonatal infection.

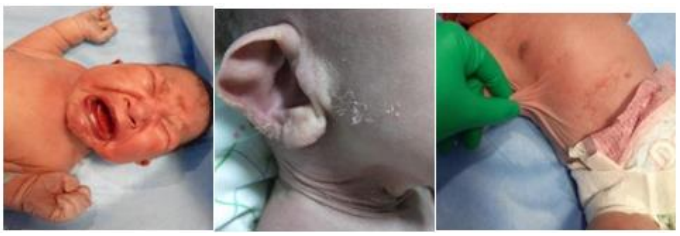

Figure1: The clinical aspect of skin laxity

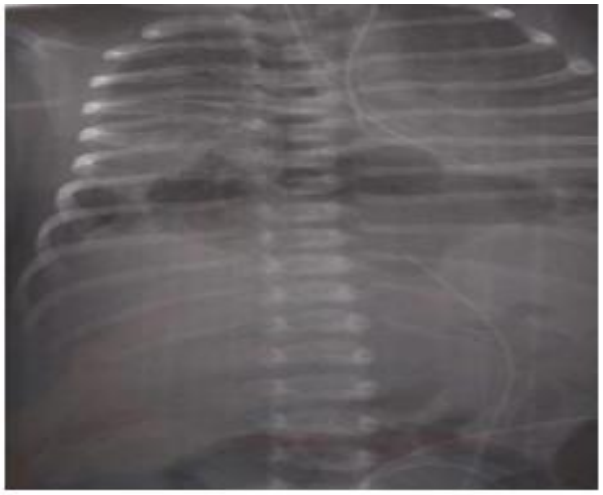

Figure2: hiatus hernia appearance on chest x-ray.

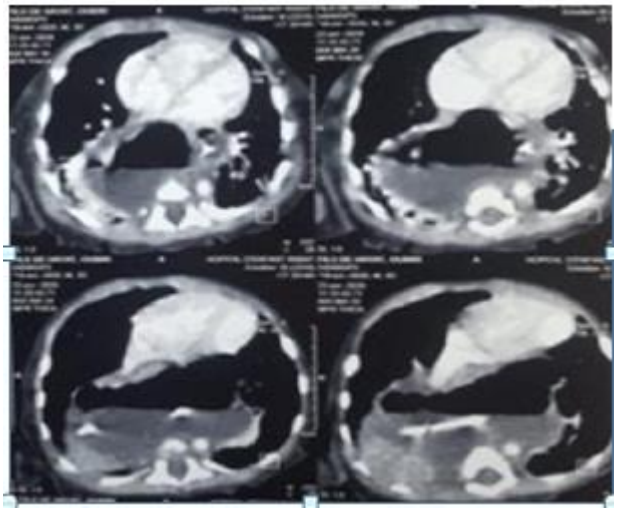

Figure3: hiatus hernia appearance on CT.

Discussion

Cutis Laxa is a rare pathology of the elastic tissue, characterized by a state of elasticity and skin slackening secondary to alterations of the elastic tissue, the elastic fibers are diminished, shortened ; The etiopathogenesis is poorly understood however a genetic mutation is implicated for the hereditary form (1). It is a condition that can be present at birth or appear in the early years of life. There are three clinical forms depending on the mode of transmission: The autosomal dominant forms, The autosomal recessive and the X-linked form (2,3). In X-linked Cutis Laxa the skin is lax, with mild mental retardation, joint laxity, and bone abnormalities. (hooked nose, pigeon chest, funnel chest), with urinary problems

and Lysyl oxidasedeficiency. The autosomal dominant form next to the lax, hanging skin, it is characterized by premature aging and pulmonary emphysema (4). The absence of visceral involvement during this form explains his better prognosis. The genetic abnormality responsible is a mutation of a gene (elastingene ELN) (2,5). While the autosomal recessive form of Type I, is the most serious form, it associates growth retardation with visceral involvement, diverticula in the esophagus, duodenum and bladder, the joints are lax and/or dislocated, the arteries are sinuous, hernias, pulmonary emphysema that define the prognosis, and responsible for deaths within the first two years in the chronic pulmonary heart disease(5), The gene whose mutation is responsible for this form has recently been identified as fibulin 5genes located on chromosome 14q31(FBLN 5).It is a gene encoding the synthesis of a protein (fibulin-5 protein) which plays an important role in the development of elastic fibers(1). Autosomal Recessive Cutis Laxa Type II Skin is Lax, with bone abnormalities, and late closure of the fontanel, associated in some cases with dislocations of the hip, the spine is curved, and the feet are flat, with an excessive tendency to dental caries. While the acquired form, later in childhood can occur either after a severe fever episode, inflammation and severe rash (Erythema Multiforme), or by an autoimmune disease or maternal medication for neonatal forms (6). There is no cure for the causes of the disease. Only the associated symptoms can be treated according to the usual protocols. It is advisable to have a "referring" doctor, close to the patient, who centralizes all the examinations as well as the opinions of the specialists in order to ensure the best possible follow-up on a daily basis(7).

The presence of hiatus hernia in cutis laxa is frequently observed in the autosomal recessive form(8), which is the case of our patient, whose clinical picture associates skin laxity with visceral involvement, which is the prognostic factor in these patients, in our case the pulmonary emphysema is not described, but his prognosis was pulmonary due to the massive hiatus hernia he presented, the mutation of the gene responsible for this form is identified under the name of fibulin 5gene, our genetic study is ongoing.

Conclusion

The inelasticity of the connective tissue is particularly visible in the skin of the body and face, when the internal organs are affected the connective tissue of these undergoes serious dysfunctions this conditions the severe vital prognosis of his children. The association described in our patient corresponds to the autosomal recessive form, which can associate the skin involvement to the serious multi-organ involvement responsible for the fatal outcomes.

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