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Cutis Laxa: A Rare Cause of Neonatal Functional Occlusion

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Abstract

Cutis laxa is a rare disease, related to loss of skin elasticity, which can be hereditary or acquired, with or without associated visceral damage. It is marked by great psychological and social repercussions. The purpose of this paper is to highlight a particular cause of neonatal occlusion: cutis laxa. We report a new observation about a case of cutis laxa hospitalized in the pediatric department at Mohammed V hospital in Tangier, admitted right after birth for the management of macrosomia with wrinkled and inelastic skin, suggesting the diagnosis of cutis laxa. The evolution that followed was marked by the occurrence of several occlusive episodes of a functional nature. Conclusion: visceral involvement in the cutis laxa is reported in several reviews. In our patient the neonatal occlusion was most likely related to her disease. The management of the case must be multidisciplinary.

Keywords

Skin Elasticity, Aging, Multisystemic Disease, Psychological Impact

1. Introduction

Cutis laxa or cutaneous hyperlaxity syndrome is a rare disorder of the elastic tissue, the etiology of which is still poorly understood It affects the skin in particular and it is well marked at the level of the face, giving an aspect of cutaneous ageing, with a great aesthetic inconvenience, causing psychological and social repercussions. However, the severity is especially related to the damage associated to it, affecting the bones or even the visceral, pulmonary in particular, vascular, gastrointestinal or urogenital parts in the form of diverticuli [1] [2].

In our patient, the visceral damage was digestive, revealed by repeated occlusive syndromes, primarily suggesting the loss of colon elasticity in relation to the

same skin disease.

The purpose of our work is to draw the practitioner's attention to the etiology of occlusions, especially when one is faced with a particular terrain.

2. Clinical Case

This is a newborn female, resulting from a 3rd act 3rd part pregnancy, that was not monitored It is said to be full term, of non-consanguineous parents, free of any affection, with no notion of a similar case in the family.

The delivery was carried out by cesarean section for fetopelvic disproportion, admitted at the 5th hour of life for the management of macrosomia. On general examination, the newborn is macrosomic with a weight of 5500 g (>97 percentile), has a height of 52 cm (>90 percentile) and a macrocranium with a head circumference of 32 cm (>3 percentile).

She is pale and hypotonic, but reacts to stimuli. Her skin temperature is normal at 37˚C. Her cardio-respiratory examination presents no particularity with a normal breathing rate of 44 cycles per minute, and a normal heart rate of 110 beats per minute.

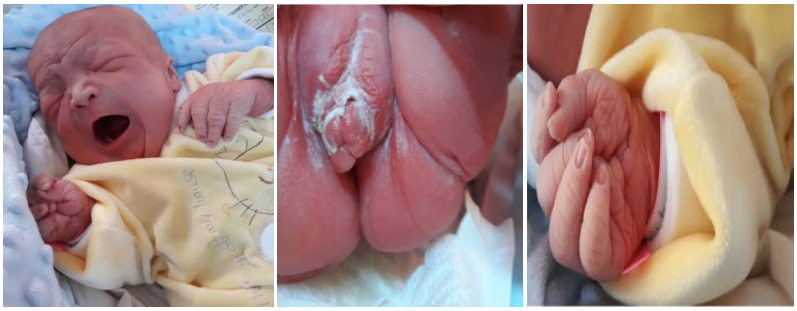
At the cutaneous-mucosal examination we note an abundant wrinkled skin hanging, forming flaccid folds giving an air of early senility. The proctological examination showed hypertrophied labia minora with clitoris and an inverted anus (**Figure 1**). The examination of the other apparatuses is without anomalies.

Monitoring during the hospitalization did not note the occurrence of hypoglycemia. Meconium was emitted at the sixteenth hour of life, the evolution was marked at 23rd hour of life by the appearance of bilious vomiting. The examination finds a distended abdomen. Abdomen without preparation showed colic hydro-aeric levels suggestive of intestinal occlusion (**Figure 2**). A supplement by abdominal ultrasound shows minimal peritoneal effusion.

Initial infection assessment and ionogram returned are correct.

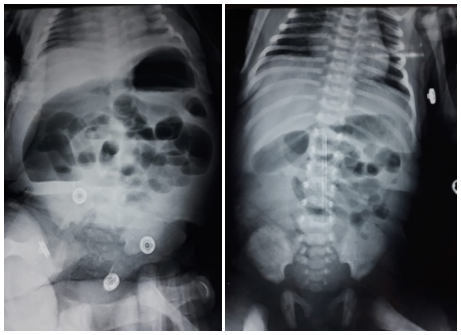
The diagnosis of neonatal occlusion probably of functional origin related to the cutis laxa was maintained. The patient is put under symptomatic treatment : a digestive rest, a basic diet through 10% serum glucose, gastric and rectal tubes with good resolution.

The medium-term evolution was marked by the recurrence of occlusive episodes with two other hospitalizations. On the 26th day of life, the baby was readmitted with the same clinical picture, the confirmation of the occlusion was obtained through the abdomen without preparation, and she was also put under symptomatic treatment with improvement of the symptomatology. On the 42nd day of life, the patient was hospitalized again with peritonitis and colonic occlusion complicated by acute dehydration. Management was urgent and aimed at initial resuscitation measures, including good rehydration with correction of hydroelectrolytic disorders. However, the patient died after two hours of hospitalization before surgery, given the delay in consultation and the severity of the clinical picture on admission.



(a) (b) (c)

Figure 1. Appearance of congenital cutis laxa in a newborn. (a) On the face. (b) External genitalia. (c) The hand.



(a) (b)

Figure 2. Thoracoabdominal Xrays. (a) Hydro-aeric levels in the colon evoking an occlusion. (b) Disappearance of the hydro-aeric levels after treatment.

In addition, the genetic study of cutis laxa was requested but not done by the family, given their lack of means.

3. Discussion

Cutis laxa is a rare heterogeneous condition, estimated to represent less than 1000 cases reported worldwide. It corresponds to a form of skin aging, which may be hereditary, or acquired secondary to inflammatory or infectious pathologies. In hereditary forms, as in the case of our patient, cutis laxa is the consequence of an abnormal metabolism of elastin linked to mutations in the elastin and fibulin genes which leads to the production of fragmented elastin, and consequently the loss of skin elasticity. At the molecular level, several genes have been implicated: FBLN5, EFEMP2 and LTBP4 in autosomal recessive type 1 cutis laxa, ATP6V0A2 and PYCR1 in autosomal recessive type 2 cutis laxa, and ELN and FBLN5 in autosomal dominant cutis laxa.

Homozygous mutations of the ELN gene have also been identified in patients with mild autosomal recessive cutis laxa.

Congenital forms can be autosomal dominant, X-linked or recessive. X-related forms are related to copper transport abnormalities and are good prognosis. Autosomal dominant forms appear late and have a relatively mild prognosis, as vis-

ceral involvement is usually moderate [1], recessive forms have different prognoses: type 1 is the most nasty for its visceral damage, especially lung [2], type 2 spares the face and laxity predominates in palmoplantar region.

Homozygous mutations in the ELN gene have also been identified in patients with a mild form of autosomal recessive cutis laxa. Congenital forms can be autosomal dominant, X-linked or recessive. The X-linked forms are related to copper transport abnormalities and have a good prognosis, the autosomal dominant forms appear late and have a relatively benign prognosis, due to the fact that visceral involvement is usually moderate [1], the recessive forms have different prognoses: type 1 is the nastiest because of its visceral involvement, especially pulmonary [2], type 2 spares the face and the laxity predominates in the palmaroplantar region.

Acquired cutis laxa are more common. Several diseases can be incriminated such as monoclonal gammopathies, lupus, acute febrile neutrophilic dermatosis, sarcoidosis, rheumatoid arthritis, infections, or drugs. They can be divided into two groups according to clinical and histological characteristics : type I which is exceptional in children, is characterized by the presence of systemic manifestations in particular cardiovascular, pulmonary and urogenital, and type II which corresponds to Marshall syndrome [3], which has two phases: an eruptive phase with papular or urticarial lesions, followed by an elastolysis phase marked by a sudden destruction of the elastic tissue in the region affected by the cutaneous damage. On the pathophysiological level, several hypotheses can be considered to explain this syndrome. Researchers incriminate the neutrophilic infiltration which can fragment the elastic fibers by mechanisms still poorly identified. Elastin alterations are the consequence of an imbalance between elastases and elastase inhibitors, responsible for pathological enzymatic digestion of the constituents of the elastic fiber and of elastin in particular.

Clinically, all forms have in common a loose, abundant, inelastic skin, with an aged appearance, and histologically a rarefaction and disorganization of the elastic fibers of the dermis with an absence of mature fibers [4].

In other words, it is the extracutaneous systemic manifestations, particularly visceral, associated with cutis laxa, which define the evolution and possibly engage the vital prognosis this implies searching for them in a systematic way.

The most frequent visceral manifestations are pulmonary and aortic damage with possible severe respiratory and hemodynamic repercussions. Musculoskeletal and digestive manifestations may be present: inguinal and umbilical hernias, intestinal or esophageal diverticula, rectal prolapse. In our patient, digestive damage is the main extracutaneous manifestation at the moment of diagnosis. Moreover, it is the reason for the hospitalization and death of the patient.

Concerning lung damage, it is most often a panlobular type of pulmonary emphysema, with neonatal respiratory distress, which occurs early or later in infancy. Performing a chest X-ray and subsequent monitoring of the cardiorespiratory state is essential, because of its evolving nature. It is the leading cause of early death. In this manifestation, the pulmonary emphysema clearly demon-

strates the crucial role of the disruption of the composition of the elastic tissue [5]. In our patient the lung was intact.

Cardiovascular damage can be primary or secondary to respiratory damage. Several malformations can be found: aortic coarctation, stenosis of the pulmonary artery, aortic or mitral valve disease, aortic ectasia, interventricular or interauricular communication [6]. Monitoring of the cardiovascular state is essential in all cases, given the evolving nature of certain manifestations [7].

Currently there is no etiological treatment, the management is initially symptomatic based on the treatment of skin manifestations by plastic surgery of skin aging, injections of botulinum toxin or filling by injection of autologous fat, especialy in the cervicofacial region where the psychological impact is more marked.

The long-term results vary according to the authors [8] [9] [10] [11]. Healing is good but the final aesthetic result is not very satisfactory due to the absence of skin retraction and the atrophic appearance of the skin, hence the need to explain these limitations to the patient.

Thus, the search for and treatment of extracutaneous manifestations is essential, such as aortic aneurysms, for which early cardiac and vascular surgery is essential.

The identification of the mode of transmission, if possible, may allow genetic counseling [8].

4. Conclusion

Cutis laxa presents a genetic and clinical polymorphism, and a rare cause of functional occlusion to be underlined. The management is multidisciplinary using plastic surgery, visceral surgery, psychological support and especially genetic counseling if the identification of the transmission mode is carried out [7].

Consent of Parents

Consent to the publication of this report has been obtained.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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