Case Report

Congenital generalized cutis laxa in two sisters

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ABSTRACT

Two sisters, aged 2 and 3 years, having generalized cutis laxa, presenting with progressive loose pendulous skin since birth are reported.

KEY WORDS: Cutis laxa, Congenital

INTRODUCTION

Cutis laxa, also called dermatochalasis or generalized elastolysis, is a heterogeneous group of elastic tissue disorders characterized clinically by lax, loose and pendulous skin resulting in a prematurely aged look.1 The condition may present as congenital2 or acquired3 forms. Involvement of the skin is usually generalized, but rarely it may be localized. The exact etiology is not known, although a disruption of the balance between elastase inhibitor and elastase in addition to a reduced production of elastin by fibroblasts has been reported.4

CASE REPORT

A 3-year-old girl born of a non-consanguineous marriage presented to us with a history of progressive, loose, lax, pendulous skin resulting in a prematurely aged appearance. At birth, the parents had noticed loose, pendulous skin of the face and the flexures, which later involved the whole body.

The child was born full term with an uneventful antenatal and perinatal period. She had delayed speech and started speaking monosyllables only at the age of three years. The other developmental milestones were normal. There was no history of dyspnea, gastrointestinal or urinary complaints. A pediatric and otolaryngology examination revealed no hearing loss or any other abnormality.

Cutaneous examination revealed loose, pendulous skin hanging in folds on the neck, chest and back, deeper nasolabial grooves, sagging cheeks and lax eyelids giving an aged look (Figure 1). Elastic recoil on pulling the skin was slow but there was no other color change in the skin. There was no laxity of the joints. The general physical and systemic examination revealed no abnormality.

Histopathological examination of skin from the back revealed that there was a decrease in number as well as granular degeneration of the elastic fibers in the dermis. The hemogram, urine examination, and liver and kidney function tests were within normal limits. A skiagram of the chest and an electrocardiogram...
revealed no abnormality.

The patient had two other siblings, an elder brother aged 5 years who was normal, and a 2-year-old younger sister who was affected similarly. The sister’s loose, lax, skin was noticed by the parents at birth, and has gradually progressed. She did not have any other systemic complaints and her developmental milestones have been normal.

DISCUSSION

Cutis laxa is a rare group of disorders in which the skin is lax and hangs in loose folds, particularly in flexures, giving the patient a prematurely aged appearance. It is of two types: congenital and acquired. In the congenital type, the inheritance is usually autosomal recessive, although autosomal dominant inheritance has been described. Our patient seems to have this rare autosomal dominant congenital form and presented with the characteristic cutaneous findings at birth. In this variety of cutis laxa the clinical manifestations are often delayed and may present only in adulthood where it may be confused with the acquired form of the disease.

In both congenital and acquired forms, the internal organs are frequently involved due to a defect in the elastic fibers and this manifests as pulmonary emphysema, pulmonary valvular stenosis, hernias and diverticulae of the gastrointestinal and urinary tracts. Our patient did not have any of these defects. Cutis laxa has been reported in association with delayed growth and development, ligamentous laxity, widely patent anterior fontanelle and congenital dislocation of the hip. In addition, cutis laxa may present as a feature of certain rare syndromes like de’Barsy syndrome where retardation of psychomotor development and corneal clouding is seen. Our patient had delayed development of speech, but no ophthalmological abnormality was detected.

Apart from being a rare skin condition, the case is being reported for its typical presentation at birth and involvement of another sibling which is not commonly reported.

REFERENCES