CONGENITAL CUTIS LAXA: A CASE REPORT

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ABSTRACT

An infant with congenital cutis laxa, a rare generalized disorder of connective tissue, is described. Radiographic manifestations were multiple diverticula of the urinary bladder, and a hiatal hernia involving the entire stomach, the right side of the colon and a distal portion of the small bowel.

INTRODUCTION

Congenital cutis laxa is an inherited disorder of connective tissue. At birth, there is a striking laxity of the skin, which hangs in pendulous folds all over the entire body. This may be especially pronounced on the face, giving an appearance of premature aging. While it is the changes in the skin which give cutis laxa its name, other manifestations of the disorder involve the pulmonary, cardiovascular, gastrointestinal, and genitourinary systems. This report describes an infant with cutis laxa, hiatal hernia and bladder diverticula.

CASE REPORT

A 6-month old boy appeared with congenitally lax skin. He was the second child, born by cesarean section at another hospital. His birth weight was 4100 grams. After birth, he had neonatal jaundice, which disappeared after 2 days of phototherapy. When he was 3 months old, he went to a private clinic because of abdominal pain, where he was advised to have a chromosome study because of his bizarre face. His older sibling looked normal. His growth and development were normal. On physical examination, the patient had generalized lax skin (Figs. 1 and 2). He also had a fever; his body temperature was 40.2 degrees Celsius. Everything else was normal. His chest film (Fig. 3) showed an air-fluid level in the right lower hemithorax and abnormal air density superimposed on the mediastinum, causing deviation of the mediastinum to the left. A barium study was done because diaphragmatic hernia was suspected. It showed herniation of the stomach into the right lower hemithorax and herniation of a loop of the large bowel into the thoracic cavity (Figs. 4 and 5). His urine had 7-8 white blood cells per high power field. *Klebsiella Pneumoniae* grew in a urine culture. After this he was given Bactrim for urinary tract infection. Then a voiding cystourethrography was done which showed multiple bladder diverticula (Fig. 6). Next the patient underwent an explore laparotomy, which showed a hiatal defect about 3 cm in diameter with paraesophageal hernia. The whole stomach, the right side of the colon, and a distal portion of the small bowel had herniated into the thorax. The surgeon reduced the visceral organs back into the abdominal cavity and closed the hiatal defect. Five days after surgery, the patient underwent an explore laparotomy again because of a recurrent hernia. At this time, the surgeon reduced the stomach back into the abdominal cavity and performed an anterior gastropexy, suturing the stomach to the diaphragm and abdominal wall. The patient has been well since.

DISCUSSION

Cutis laxa is a rare disorder of connective tissue in which there is reduction in the amount and size

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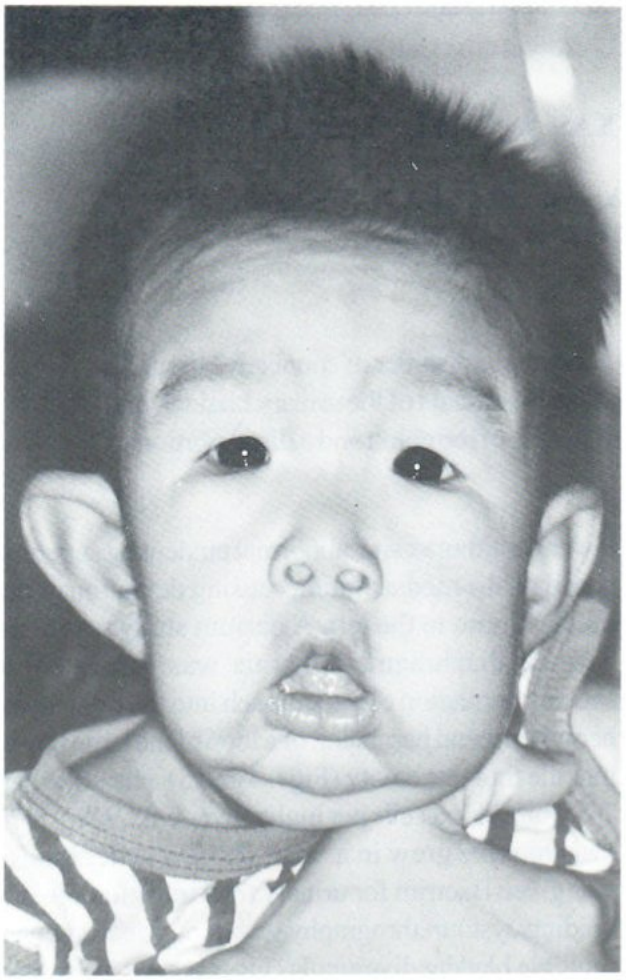

Fig.1 Strikingly elderly looking face and huge ears.

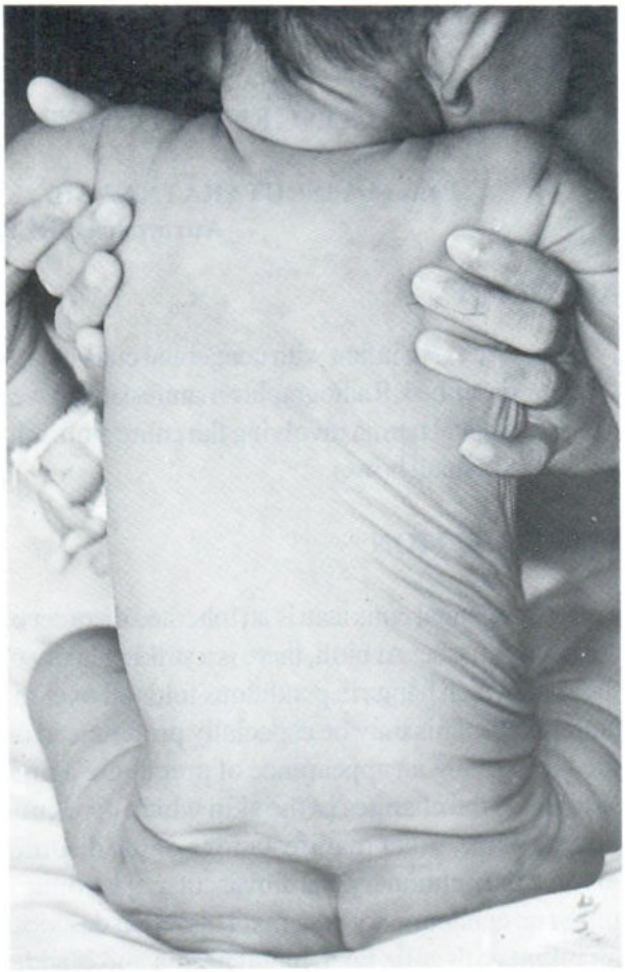

Fig.2 Generalized lax skin is evident in many loose olds over back, buttocks, and thighs.

of the elastic fibers.[1] The etiology is unknown. Both congenital and acquired forms have been described .[2,3,4] Modes of inheritance include autosomal dominance with variable penetrance, autosomal recessiveness present at birth or shortly after birth and X-linked recessiveness.[1] Though time of onset of symptoms and clinical presentation may vary in these patients, this syndrome is always characterized by loose, pendulous, inelastic folds of skin.

The reported radiologic manifestations include pulmonary emphysema,[2] hernia,[3,5] dilatation and tortuousity of blood vessels,[5] and diverticula of the gastrointestinal and urogenital tracts.[4,5]

In this case, the hiatal hernia which recurred after the first surgery, the multiple bladder diverticula; and the generalized lax skin were the results of abnormality of elastic fibers. The roentgenographic findings of hiatal hernia and multiple bladder diverticula described in this patient can also be seen in patients with Ehlers-Danlos syndrome; however, hyperelasticity of the skin, hyperextensible joints, bleeding diathesis and soft-tissue calcifications characteristic of Ehlers-Danlos syndrome[6] were lacking in this patient.

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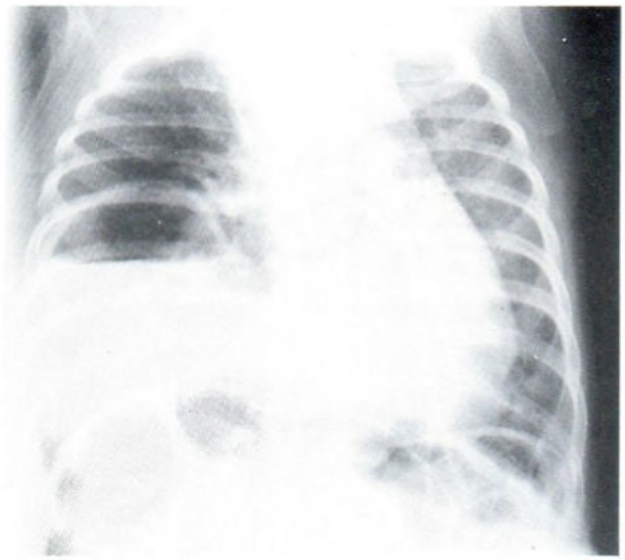

Fig.3 Chest film shows an air-fluid level in the right lower hemithorax. Abnormal air density is also seen superimposed on the mediastinum, and causing it to deviate to the left.

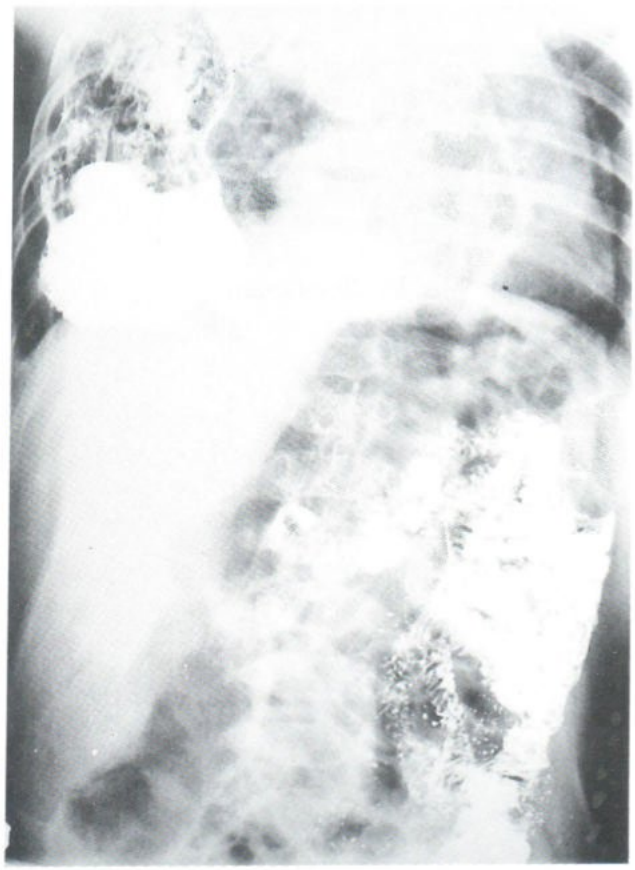

Fig.4 Barium study shows herniation of the stomach into the right lower hemithorax.

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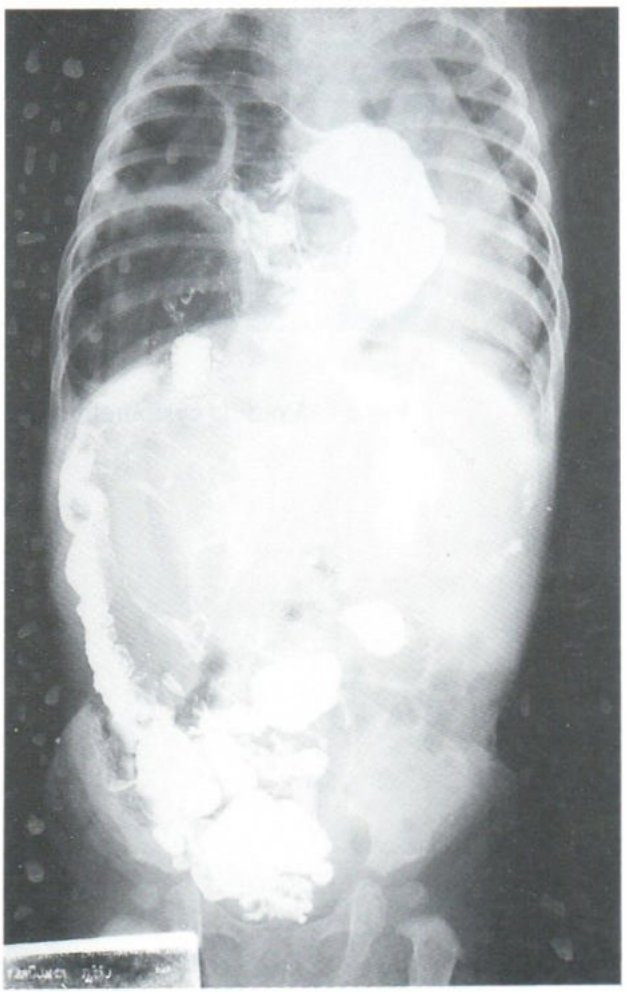

Fig.5 Barium study at 10 hours shows herniation of a portion of large bowel into the thorax.

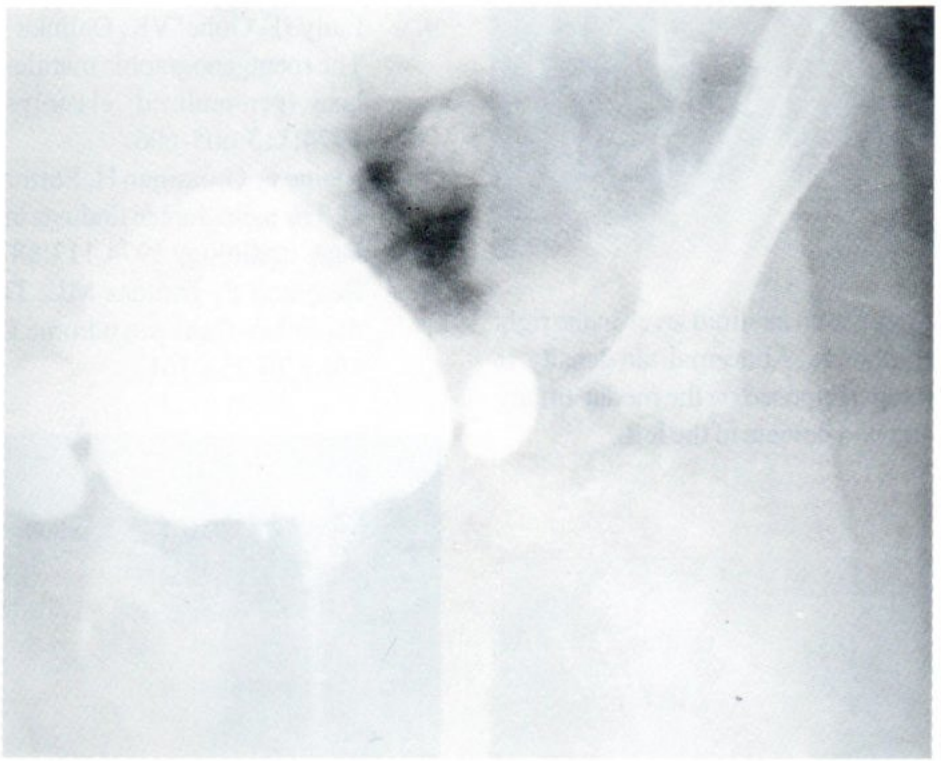

Fig.6 Voiding cystourethrogram shows multiple bladder diverticula.