

## CASE REPORT ΕΝΔΙΑΦΕΡΟΥΣΑ ΠΕΡΙΠΤΩΣΗ

### Cutis laxa Case report

Cutis laxa is a rare syndrome due to a defect in the production and destruction of elastic tissue, which is characterized by loose skin, wrinkled skin and premature skin aging. The case is presented of a female with clinical manifestations suggestive of cutis laxa syndrome with a twin brother who died with similar clinical features. The disorder may be transmitted by autosomal dominant and autosomal recessive mode, the latter usually having more severe manifestations. For the definitive diagnosis and subtyping of the syndrome genetic study should be performed.

Cutis laxa is a rare syndrome which is a genetic disorder causing elastic tissue dysfunction.<sup>1-3</sup> In this disease, disturbance in the network of elastic fibers results in premature aging of the skin.<sup>4,5</sup>

The inheritance of the disease is of two types, autosomal dominant and autosomal recessive. The autosomal dominant type has milder manifestations, and the patients usually have a normal life, with delayed appearance of symptoms.

The recessive type is usually diagnosed at birth or even in the fetal stage. In the autosomal recessive type other manifestations, including inguinal hernia, pulmonary emphysema, vascular problems and digestive disorders may be present and this type has a high mortality.<sup>3,6-8</sup>

#### CASE PRESENTATION

The case is reported of a newborn girl, the first child of a twin pregnancy born at 36 weeks gestation. The second twin of this pregnancy, a male, had a cephalic presentation and Apgar score 8 and intrauterine growth retardation (IUGR) and did not survive.

The first twin was born with Apgar score 8, birth weight 2,985 grams and respiratory distress. On examination, the patient had a short neck and the skin of her face was pendular with sagging folds (fig. 1). On auscultation a heart murmur was detected at

ARCHIVES OF HELLENIC MEDICINE 2017, 34(2):253-254  
ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2017, 34(2):253-254

**B. Darban,  
M. Moradi**

*Pediatric Department, Hormozgan  
University of Medical Sciences, Bandar  
Abbas, Iran*

Χαλαροδερμία: Περιγραφή  
περίπτωσης

*Περίληψη στο τέλος του άρθρου*

#### Key words

Cutis laxa  
Genetic study  
Wrinkled skin

*Submitted 6.6.2016  
Accepted 19.7.2016*



**Figure 1.** Female neonate with cutis laxa syndrome: Loose skin of the body, with sagging folds.

the LSB. Echocardiography showed moderate aortic dilatation.

On laboratory testing, the patient had a normal complete blood count, normal BUN, creatinine and electrolytes, and efficient G6PD. Total bilirubin was 11 mg/dL and direct bilirubin was 0.5 mg/dL.

The diagnosis of cutis laxa was made on clinical evidence, as there was no access to genetic testing of the patient, and due to lack of parental permission, skin biopsy was not performed.

## DISCUSSION

Cutis laxa syndrome is a connective tissue disorder that occurs due to a defect in the production and destruction of elastic fibers, which present as rubbery stems. The main

manifestations include skin involvement, consisting of loose skin, wrinkled skin, and premature skin aging.<sup>1-4</sup>

Systemic manifestations include pulmonary emphysema, large vessel aneurysms, inguinal and umbilical hernia, and gastrointestinal diverticulum.<sup>2,3,5,6</sup>

The disorder may be transmitted by autosomal dominant or autosomal recessive mode. For the definitive diagnosis and subtyping of the syndrome genetic study should be performed.<sup>3,8</sup>

In the past when genetic study was not possible, the diagnosis was based on physical examination and histological evaluation of skin biopsy, but now genetic analysis is required for the diagnosis.

## ΠΕΡΙΛΗΨΗ

### Χαλαροδερμία: Περιγραφή περίπτωσης

B. DARBAN, M. MORADI

*Pediatric Department, Hormozgan University of Medical Sciences, Bandar Abbas, Ιράν*

*Αρχεία Ελληνικής Ιατρικής 2017, 34(2):253-254*

Η χαλαροδερμία είναι ένα σπάνιο σύνδρομο οφειλόμενο σε ανεπάρκεια παραγωγής και καταστροφή του ελαστικού ιστού, το οποίο χαρακτηρίζεται από χαλαρό και ρυτιδωμένο δέρμα καθώς και από παρουσία πρόωρης γήρανσής του. Παρουσιάζεται η περίπτωση ενός θήλεος βρέφους με κλινικές εκδηλώσεις που υποδηλώνουν χαλαροδερμία, ο δίδυμος αδελφός της οποίας πέθανε με παρόμοιες κλινικές εκδηλώσεις. Η διαταραχή μεταβιβάζεται με τον αυτοσωματικό επικρατή και τον αυτοσωματικό υπολειπόμενο χαρακτήρα. Για την οριστική διάγνωση και τον καθορισμό του υπότυπου του συνδρόμου απαιτείται γενετική μελέτη.

**Λέξεις ευρητηρίου:** Ρυτιδωμένο δέρμα, Χαλαρό δέρμα, Χαλαροδερμία

## References

1. AL-FAKY YH, SALIH MA, MUBARAK M, AL-RIKABI AC. Bilateral congenital entropion with cutis laxa. *Pediatr Dermatol* 2014, 31:e82-e84
2. KUMAR M, SINGH R. Congenital cutis laxa. *Indian Pediatr* 2012, 49:771
3. NASCIMENTO GM, NUNES CS, MENEGOTTO PF, RASKIN S, ALMEIDA ND. Cutis laxa: Case report. *An Bras Dermatol* 2010, 85:684-686
4. RIVEROS CJ, GAVILÁN MF, FRANÇA LF, SOTTO MN, TAKAHASHI MD. Acquired localized cutis laxa confined to the face: Case report and review of the literature. *Int J Dermatol* 2004, 43:931-935
5. WEIR EK, JOFFE HS, BLAUFUSS AH, BEIGHTON P. Cardiovascular abnormalities in cutis laxa. *Eur J Cardiol* 1977, 5:255-261
6. GUÍA TORRENT JM, CASTRO GARCÍA F, CUENCA GÓMEZ M, GRACIÁN GÓMEZ M. Cardiovascular changes in the cutis laxa congenita syndrome. *Rev Esp Cardiol* 1999, 52:204-206
7. GIRISGEN I, MUNGAN I, CAKIR M, ERSOZ S, OKTEN A. Autosomal recessive cutis laxa in two siblings associated with blue sclera. *Pediatr Dermatol* 2007, 24:E10-E13
8. TANTCHEVA-POOR I, SCHUSTER A, KORNAK U, CHELIUS K, MAUCH C. Congenital autosomal recessive cutis laxa type II. A wrinkly-skin-syndrome. *Klin Pädiatr* 2012, 224:322-323

*Corresponding author:*

B. Darban, Pediatric Department, Hormozgan University of Medical Sciences, Bandar Abbas, Iran  
e-mail: darban.sam@gmail.com