CASE REPORT

A Case with Cutis Laxa Syndrome which is Bilateral Developmental Dysplasia of the Hip

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ABSTRACT

Cutis laxa syndrome (CLS), known as generalized elastosis, is a rarely occurring connective tissue disease, showing up in two separate ways, congenital, or acquired. It makes skin to lose elasticity and folds sagged. In this study, a case with CLS has been reported, which is bilateral developmental dysplasia of the hip.

Key words: Developmental dysplasia of the hip, elastin, cutix laxa

INTRODUCTION

The exact cause of cutis laxa syndrome (CLS) is still not certain, but some probable causes of this disease are reduced dermal elastin, content depending on abnormal elastin metabolism, and degradation of the elastic structure of the skin.[1,2] CLS can be seen as acquired CLS which is rare, most cases are congenital CLS.[3]

Patients with CLS have a characteristic face appearance and CLS reveals with the symptoms such as front-facing nostrils and lower eyelid rim facing downward. Depending on the loose in the vocal cords, it can also make voice crying. The degree of involvement may vary from regional skin, widespread skin to systemic involvement (pulmonary, vascular, cardiac, gastrointestinal, and genitourinary).[4-6]

The aim of this study is to discuss a case with CLS which has bilateral developmental dysplasia of the hip and to present it in the light of literature.

CASE REPORT

A 7-year-old girl applied to our hospital, suffering from the appearance of her skin and also has a delay while walking. She was born as the third child of a healthy, relative couple, and born through the normal vaginal way in the right time. Physical examination of the patient revealed good general condition and her consciousness was clear. There was no elasticity increase on the wrinkly skin, and no pathological symptom detected on respiratory and cardiovascular examination. The abdomen was in the wrinkled appearance and had no hepatosplenomegaly and hernias [Figure 1]. Reflexes evaluated as normal in the neuromuscular examination. She was a normal female patient, except her urogenital system. Extremities were symmetric but the skin on the extremities was wrinkled. There were incision scars on both lateral tights caused by bilateral hip reduction operations. No purpura, petechiae, and ecchymosis determined. On linkages, there was no increase in elasticity and mobility. The patient had Trendelenburg gait, increased lumbar lordosis, and limited range of abduction. Bilateral hip dislocation was determined with radiography, and bilateral radical reduction operation was performed. The patient was discharged without any complications at intraoperative and post-operative time [Figures 2 and 3].

DISCUSSION

Congenital CLS is a genetical syndrome which is inherited as autosomal dominant and recessive form.[7] Autosomal dominant form is more benign. Parents of the patient with

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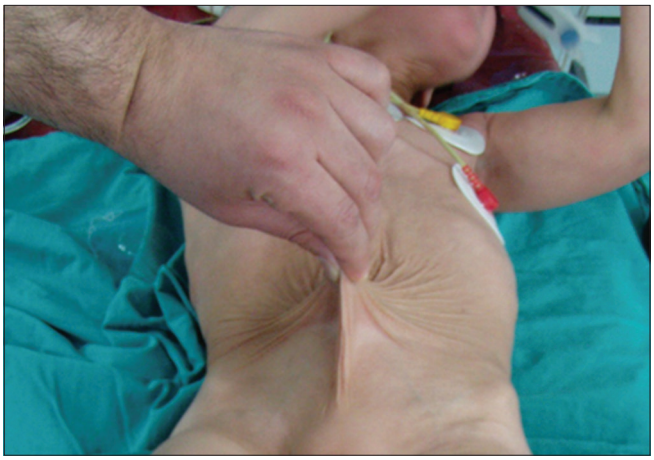

Figure 1: Wrinkly skin appearance patient with cutix laxa syndrome

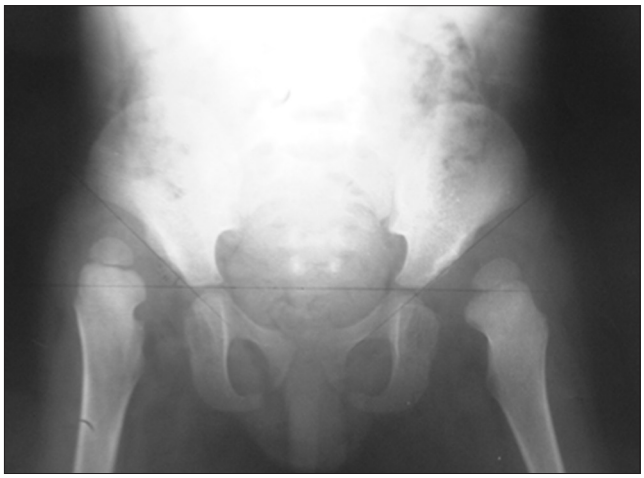

Figure 2: Bilateral developmental dysplasia of the hip of the patient's preop X-ray

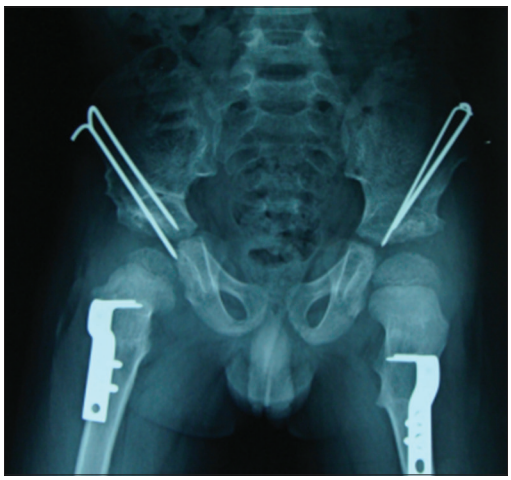

Figure 3: Bilateral developmental dysplasia of the hip of the patient's post-operative X-ray

autosomal dominant CLS had no disease history, and it arises with gene mutation on the elastin gene.[8-10]

The etiopathogenesis of the disease has not been fully clarified. Formerly, its cause asserted due to low serum copper levels and high urinary copper extraction. Low copper levels cause lack of (xx) which serves as elastase inhibitory and lack of (xx) causes more destructed elastic fibers.[11] Histopathologic findings are basically loss of elastic tissue in the dermis. Abnormal fragmentation on amorphous elastin component, a decrease in the dermis elastin microfibril along with a decrease in the presence of elastic fibers, has been shown with electron microscopic examination in the skin biopsy.[12,13] Elastic fibrils consisted of two components: Amorphous component (elastin) which is more common and microfibriler component and elastin substantially comprised glycine, proline, and some hydrophobic residues.

Early-onset prognosis is better than late-onset prognosis in patients.[14] Systemic elastolysis risk (bowel diverticula, inguinal and hiatal hernias, aortic rupture, emphysema, and cor pulmonale) is high in adult acquired CLS patients aged >20 years old.[15] Patients with cardiovascular and pulmonary diseases have a very high death risk (almost % 100).[16]

A total of 30 cases have been reported the association with CLS and intrauterine and postnatal growth retardation. Internal organ symptoms and growth retardation are mostly associated with the recessive form and less associated with autosomal dominant form.[17-19] Our examination revealed that our patient had no respiratory and cardiovascular problems and also no pathology has been determined in abdominal ultrasonography. In addition to sagged folds, changes in face appearance make patients appearance older, and it worsens as the time passes.[20] Delayed fontanelle closure of the cases accompanied by disease was observed in our case.

Central nervous system (CNS) mostly gets confused with Ehlers-Danlos syndrome (EDS) in the differential diagnosis. EDS symptoms are; elastic and loose skin increased elasticity and hypermobility in joints.[21]

The pathogenesis of developmental dysplasia of the hip is multifactorial due to a combination of hormonal, positional, and familial factors. Primarily some periods of *in utero* development are precarious for hip dislocation or hip dysplasia. Thus, 12 and 18 weeks of pregnancy and the past 4 weeks of gestation are important. Furthermore, some physiological factors have a role in the etiology of CNS, as hormonal and hereditary ligamentous laxity, causes bond relaxation on hip area.[22]

Our patient has been operated due to bilateral hip dislocation. In literature, it's reported that three intermarriage family with CLS had 5 hip dislocations .[23] In the contrast of previously mentioned findings of the etiopathogenesis, we believe hip dysplasia of our patient may be incidental.

REFERENCES

1. Biver A, De Rijcke S, Toppet V, Ledoux-Corbusier M, Van Maldergem L. Congenital cutis laxa with ligamentous laxity and delayed development, dandy-walker malformation and minor heart and osseous defects. Clin Genet 1994;45:318-22.
2. Lewis FM, Lewis-Jones S, Gipson M. Acquired cutis laxa with dermatitis herpetiformis and sarcoidosis. J Am Acad Dermatol 1993;29:846-8.
3. Chun SI, Yoon J. Acquired cutis laxa associated with chronic urticaria. J Am Acad Dermatol 1995;33:896-9.
4. Turner RB, Haynes HA, Granter SR, Miller DM. Acquired cutis laxa following urticarial vasculitis associated with igA myeloma. J Am Acad Dermatol 2009;60:1052-7.
5. Newlove T, Tzu J, Meehan S. Papillary dermal elastosis. Dermatol Online J 2011;17:12.
6. Larangeira de Almeida H Jr. Passos da Rocha M, Neugebauer S, Wolter M, Rocha NM. Acquired cephalic cutis laxa. Dermatol Online J 2007;13:31.
7. Beighton P. The dominant and recessive forms of cutis laxa. J Med Genet 1972;9:216-21.
8. Zhang MC, He L, Yong SL, Tiller GE, Davidson JM. Cutis laxa arising from a frame shift mutation in the elastin gene (ELN). Am J Hum Genet 1997;61 suppl: A353.
9. Zhang MC, He L, Giro M, Yong SL, Tiller GE, Davidson JM, *et al.* Cutis laxa arising from frameshift mutations in exon 30 of the elastin gene (ELN). J Biol Chem 1999;274:981-6.
10. Agha A, Sakati NO, Higginbottom MC, Jones KL Jr. Bay C, Nyhan WL, *et al.* Two forms of cutis laxa presenting in the newborn period. Acta Paediatr Scand 1978;67:775-80.
11. Harrington CR, Beswick TC, Susa JS, Pandya AG. Acquired cutis laxa associated with heavy chain deposition disease. J Am Acad Dermatol 2008;59:S99-101.
12. Lustmann J, Nahlieli O, Harary D, Casap N, Neder A, Zlotogora J, *et al.* Gerodermia osteodysplastica: Report on two patients and surgical correction of facial deformity. Am J Med Genet 1993;47:261-7.
13. Thomas WO, Moses MH, Craver RD, Galen WK. Congenital cutis laxa: A case report and review of loose skin syndromes. Ann Plast Surg 1993;30:252-6.
14. Berk DR, Bentley DD, Bayliss SJ, Lind A, Urban Z. Cutis laxa: A review. J Am Acad Dermatol 2012;66:842.e1-17.
15. Xue Y, Chen H, Zeng X, Jiang Y, Sun J. Generalized acquired cutis laxa treated with facial plastic surgery. Eur J Dermatol 2011;21:141-2.
16. Lewis KG, Bercovitch L, Dill SW, Robinson-Bostom L. Acquired disorders of elastic tissue: Part II. Decreased elastic tissue. J Am Acad Dermatol 2004;51:165-85.
17. Patton MA, Tolmie J, Ruthnum P, Bamforth S, Baraitser M, Pembrey M. Congenital cubs laxa with retardation of growth and development. J Med Genet 1987;24:556-61.
18. Sakati NO, Nyhan WL, Shear CS, Manno CS, Maris JM, Rhodin N. Syndrome of cubs laxa, ligamentous laxity and delayed development. Pediatrics 1983;72:850-6.
19. Karakurt C, Sipahi T, Ceylaner S, Senocak F, Karademir S, Becer M, *et al.* Cutis laxa with growth and developmental delay. Clin Pediatr (Phila) 2001;40:422-3.
20. Philip AG. Cutis laxa with intrauterine growth retardation and hip dislocation in a male. J Pediatr 1978;93:150-1.
21. Lewis PG, Hood AF, Barnett NK, Holbrook KA. Postinflammatory elastolysis and cutis laxa. J Am Acad Dermatol 1990;22:40-8.
22. Grissom LE, Harcke HT. Ultrasonography and developmental dysplasia of the infant hip. Curr Opin Pediatr 1999;11:66-9.
23. Karrar ZA, Elidrissy AT, Adam KA. Cutis laxa, intrauterine growth retardation, and bilateral dislocation of the hips: A report of five cases. Prog Clin Biol Res 1982;104:215-22.

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