Case Report

Autosomal recessive Cutis laxa with an inguinal hernia: a rare inherited disorder of connective tissue

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Abstract

Cutis laxa (CL) is a rare connective tissue disorder characterized by loose, sagging, and redundant skin. Both inherited and acquired forms are known. A 16-month-old boy, presented with loose, redundant skin chiefly below the chin, trunk, extremities since birth with scrotal swelling and difficulty in micturition. The patient had a characteristic senile bloodhound appearance. Histopathological examination showed markedly reduced elastic fibres in the dermis. There is no definitive treatment available at present. Differential diagnosis includes Ehlers–Danlos syndrome, congenital disorders of glycosylation syndrome, and pseudoxanthoma elasticum. That case is an important subject to report for its rarity and complex systemic involvement.

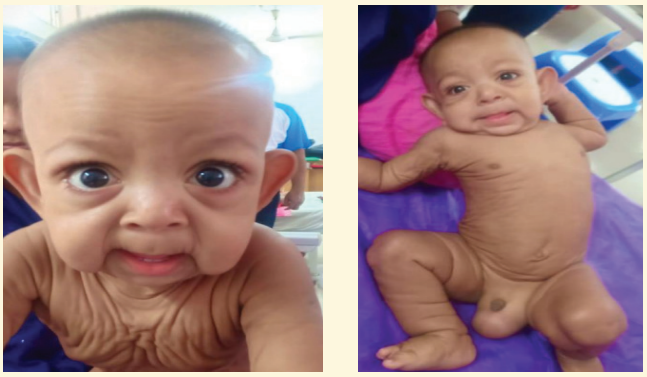
Key words: Cutis Laxa, Elastolysis, Autosomal recessive cutis laxa.

Introduction:

Cutis laxa (CL) is a rare disorder of elastic tissue. Only over 200 cases reported worldwide so far.[1] It is characterized by elastolysis or loss of elasticity of the skin. Patients usually presented with a characteristic old appearance with loose, sagging skin over the face, extremities and trunk. Both inherited and acquired forms are found. Inherited forms of CL may be autosomal dominant, autosomal recessive (AR), and X-linked.[2] Normally the disease can be diagnosed clinically with minimal laboratory support. There is no treatment for cutis laxa. Some supportive, symptomatic and or cosmetic treatment options can improve the quality of life of patients.

Case report:

A 16-month-old boy the third and only living child of a consanguineous parent presented with loose and inelastic skin over the face, trunk and extremities noticed since the 4 th month of life. The boy looked like an old man. His IQ and developmental milestones are up to the mark. The patient's mother had complaint of his difficulty in micturition and gradual swelling of the scrotum. He had no other systemic complaint



(A) (B)

Figure 1: a) A case of cutis laxa b) Scrotal swelling

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His eldest sister was a case of cutis laxa and died with pulmonary emphysema and corpulmonale when she was at the age of 16 months. His elder brother died at his first month of life due to pneumonia. He was not diagnosed as a case of cutis laxa. His imaging and the echocardiographic report were normal but ultrasonographic study confirms bilateral inguinal hernia. There was a loss of elastic tissue in the dermis with Verhoeff-Van-Gieson stain in the histopathological report. Ptosis, low set, prominent ears, large forehead, long philtrum, and hanging cheeks were present over the face, giving the child an aged appearance. He was at first diagnosed as a case of progeria might be for his characteristic facies.

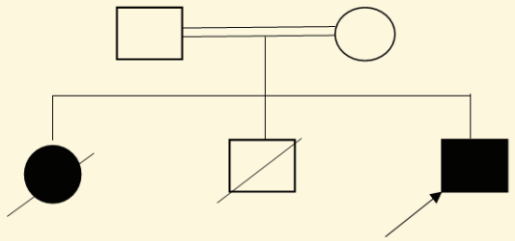

Figure 2: Pedigree chart of patient

The child was born out of a first-degree consanguineous marriage. His mother stated that her antenatal period and labour were uneventful. Finally, the patient was diagnosed as a case of autosomal recessive cutis laxa type 1 (ARCL-1). He was sent for consult with a pediatric surgeon for his hernia.

Discussion:

Generalized CL is also known as generalized elastolysis.[3] The calculated prevalence is 1/1,000,000.[4] ADCL is a milder form and usually appears in childhood or early adulthood. It presents with characteristic facial features with loose skin and systemic involvement such as gastrointestinal diverticula, herniae, uterine prolapse, and aortic dilatations.[2,4] Autosomal recessive cutis laxa (ARCL) is the most severe form of CL. ARCL-1 presents at birth with cutaneous manifestations affecting the whole body. Systemic manifestations include pulmonary emphysema, gastrointestinal and bladder diverticula, arterial aneurysms, pulmonary artery stenosis, hernia, etc., and are severe and life-threatening.[5] Child's developmental milestones and intelligence quotient are normal. FBLN5 and FBLN4/EFEMP2 genes are affected.[4] In ARCL-2, there is intrauterine growth restriction, skeletal involvement, and developmental delay. Loss of function mutations is present in ATP6V0A2, resulting in abnormal glycosylation.[2,4,6] ARCL-3 (de Barsy syndrome) presents with severe mental retardation, athetoid movements, and corneal opacities.[7] In X-linked form (occipital horn syndrome), bladder diverticula, inguinal hernias, slight skin laxity and hyperextensibility, joint laxity, coarse hair, and skeletal abnormalities are associated. Mutation in copper transporter gene ATP7A is seen, resulting in low-serum copper, and ceruloplasmin levels.[2,4] Differential diagnosis includes Ehler's–Danlos syndrome in which there are hyperextensible joints with hypermobility, but not lax skin. There is quick recoil of skin on stretching.[2,4,8] Pseudoxanthoma elasticum is characterized by fragmented and calcified elastic fibres. The face is spared, and the skin is yellow.[2,4] The exact pathogenesis of CL is not known, but the various hypothesis has been proposed such as immune-mediated mechanism, altered copper metabolism, and excess elastase.[2,4]
In our case, the child had similar complaints in siblings, which is suggestive of AR inheritance. In our case, there was a severe wide range of systemic involvement (pulmonary hypertension, emphysema, bronchopneumonia, chronic cystitis and bladder diverticula) which are life-threatening.[9] Hence, continuous monitoring and active management for the associated complications were needed. Diagnosis of ARCL-1 was done based on clinical features, extracutaneous findings, and histopathological findings with the exclusion of differential diagnosis. There is no effective treatment available for ARCL-1.[10,11] Symptomatic management for the associated complications remains the mainstay of treatment. Genetic counselling of the affected family is essential.[12] Due to financial constraints the genetic studies could not be performed in our case. It might be the first reported case of cutis laxa in Bangladesh. Conclusion: We report this case of ARCL-1 with complex systemic involvement for its rarity.

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