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CASE REPORT

Cutis Laxa in Pediatrics with Hiatal Hernia: A Rare Presentation

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ABSTRACT

Cutis laxa (CL) is a rare connective tissue disorder which may be inherited or acquired. It is characterized by a loose skin. Inherited forms are autosomal dominant, autosomal recessive and X-linked recessive. Congenital cutis laxa may present with variable systemic involvement e.g (gastrointestinal, CNS and cardiopulmonary) determining a worst prognosis. Acquired forms of this disease have been associated with a past inflammatory skin disorder. We report a 4 year old boy with a congenital cutis laxa. He was admitted for vomiting and constipation. Diagnosis of cutis laxa is typically made by physical examination and associated extra cutaneous finding. Cutis laxa is a rare disorder, diagnosed clinically although there is wide clinical and genetic heterogeneity.

Key Words: *Cutis laxa, Connective tissue disorder, Loose skin.*

INTRODUCTION

Cutis laxa is a heterogeneous connective tissue disorder the disease is quite rare and identified by premature ageing of the skin.[1,2] Both inherited and acquired forms exist. Congenital form is most common. Autosomal dominant form also exists.[3] The most obvious symptom of cutis laxa is loose wrinkled skin, especially around the face, trunk, arms, and legs, which hangs in folds and causes an aged appearance. Other systemic manifestation include GIT, CVS, CNS and respiratory system. The disease is caused by paucity of elastic fibers. Patients with the autosomal recessive type have a high risk of serious cardiopulmonary and gastrointestinal complications.[4] And any extra-cutaneous manifestation is a bad prognostic factor.

CASE REPORT

IG, a 4 year old boy, born to consanguineous parents (2nd degree relative), presented with complaints of vomiting and constipation. There was history of vomiting multiple episodes associated with food intake and constipation for 10 days. He was receiving treatment from local doctors due to on off respiratory distress, and was referred to us for further workup and management. There was history of similar disease in one paternal female cousin who was 7 year old. The child was developmentally normal, afebrile, conscious and cooperative with no obvious respiratory distress. The vitals were: RR 26/min, HR 76/min, and blood pressure 90/70 mmHg, SpO_2 was 98%. The weight and height were 11 kg and 94 cm (both under 2SD) respectively. The face had lax skin hanging in folds prominent around eyes (fig 1A). Palate was high arched. Eyes were normal. His thorax was normal in shape and abdomen was sunken. There was a bilateral, reducible, inguinal hernia (fig 1B). Joints examination revealed excessive mobility more prominent around wrist joint, also involving elbow joint. Chest examination was normal. CVS exam revealed holosystolic murmur at left lower sternal area with loud P2, over pulmonary area. There

was no palpable liver or spleen. Others systems were normal. Test showed Hb: 12.40 g/dl, TLC: 7700/mm3 with neutrophil 51% and lymphocytes 42%. Routine biochemical investigations were normal. Patient also had subclinical hypothyroidism with raised TSH: 5.4 microlU/ml free T32.3 pg/ml and T41.2 ng/dl. CXR was normal. Echocardiography showed mild tricuspid regurgitation, mild to moderate pulmonary arterial hypertension and otherwise structurally normal heart. USG abdomen was normal. Barium studies done revealed hiatal hernia with gastro-esophageal reflux and possible mesentroaxial gastric volvulus showing proximal contrast hold up and contrast trickling opacifying small bowel loops (fig 2). The child was treated with antiemetics. And referred to pediatric surgery for operative procedure where gastropexy, resection of esophageal stricture with end to end anastomosis was done.

Fig 1 (A): Senile appearance of the face

Fig 1 (B): Bilateral inguinal hernia

Fig 2: barium study showing hiata hernia and gastric vovolus

DISCUSSION

Cutis laxa is a rare connective tissue disorder recognized clinically by loosely hanging skin. It is caused by reduction in number or altered structure of elastic fibers in skin. Autosomal recessive type has severest clinical manifestations. Children with this disease have loose skin at birth, and if cardiopulmonary problems appear early than survival beyond infancy is difficult. Systemic manifestations include large vessel aneurysms, pulmonary emphysema, pulmonary stenosis, hernia, and diverticula in the gastrointestinal and genitourinary tracts. It is caused by mutation in fibrillin gene.[4] While autosomal dominant form is a mild condition without other organ involvement, caused by mutations in the elastin gene.[5]

The third type of cutis laxa is due to X-linked ATP7A gene. It is also known as occipital horn syndrome and shares similarities to Menke"s disease.[6] In our patient, there is history of similar problem in one female cousin, likely a case of autosomal recessive pattern. He had no history of developmental delay. He had the typical skin abnormalities observed in all the types of cutis laxa. Additionally, he had inguinal hernia, cardiac manifestation and gastro-intestinal involvement. Various GIT problems like diverticulae, stenosis, hiatal hernia as in our patient have been reported, Kothari et al reported the first neonate with cutis laxa with multiple hernias and cardiac disorder.[7] Other studies also reported neonates presenting with hiatal hernia.[8] One study reported hiatal hernia in a 50 year old female presenting with recurrent illeus.[9] But upon comprehensive search we found that our study is first to report hiatal hernia associated with volvulus in children beyond neonatal age group. Our patient also had subclinical hypothyroidism. Koklu et al was first to report hypothyroidism in cutis laxa.[10] Many other cases also reported the same.

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