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Case Report

Congenital cutis laxa with multi-system involvement

Karan Malhotra*, Karjigi Siddalingappa, Kallappa C. Herakal

Department of Dermatology, Venereology and Leprosy, Navodaya Medical College, Hospital and Research Centre, Raichur, Karnataka, India

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***Correspondence:**
Dr. Karan Malhotra,
E-mail: dr.karan2015@gmail.com

ABSTRACT

Cutis laxa is a heterogeneous group of inherited and acquired rare connective tissue disease characterized by loose, wrinkled, and inelastic skin. It clinically presents as loose skin with folds giving a premature aged appearance. Cutis laxa is very rare, with an estimated incidence of one in 4 million. There are many case reports on acquired cutis laxa but very few on "congenital" cutis laxa. Authors report a 15 years old female presenting with a history of recurrent respiratory tract infections since the age of 2 years associated with flaccid skin all over her body and extensive loose folds of skin over face, neck, abdomen, arms and thighs since birth. Cutis laxa has been diagnosed based on the clinical picture and histopathological appearance. No medical treatment is available for correction of the pathology of disease. Plastic surgery remains the only modality of treatment to improve the cosmetic appearance. Systemic abnormalities need specific treatment depending upon the condition. The purpose of this report is due to its rarity and involvement of skin, hairs, respiratory, cardiovascular and genitourinary system in a single patient.

Keywords: Connective tissue disease, Congenital, Cutis laxa, Inherited, Flaccid skin

INTRODUCTION

Cutis laxa (CL) is a heterogeneous group of inherited and acquired rare connective tissue disease characterized by a loose, wrinkled, and inelastic skin. CL can be congenital or acquired.[1] Congenital type is accompanied by systemic complications, such as pulmonary, cardiac and vascular complications.[2] Cutis laxa is very rare, with an estimated incidence of one in 4 million.[3] There are very few case reports of congenital cutis laxa.

CASE REPORT

A 15 years old female child, born to consanguineously (second degree) married parents, presented with complaints of fever, cough with expectoration and gradually progressive breathlessness from mMRC (Modified Medical Research Council) Dyspnoea scale +1 → +3 since 2 weeks. There was a history of recurrent episodes of respiratory tract infections since the age of two years. She was receiving treatment from a local practitioner but was referred for increasing breathlessness. Parents gave history of flaccidity of skin all over her body with extensive loose folds of skin over face, neck, abdomen, arms and thighs since birth. History of delayed developmental milestones with low IQ. Insignificant family history with regular menstrual cycles (3-4 days/ 28 days).

On examination, the child was febrile with dyspnoea, tachypnoea, and intercostal and subcostal retractions. Vitals: Pulse rate- 130/min, BP- 110/70 mm Hg, Respiratory rate- 45/min, Temperature: 104^0F, and SpO2 was 65% in the ambient air. The weight and height were 25 kg (<3rd percentile) and 110 cm (<3rd percentile) respectively.

Head to toe examination: Hairs were sparse and brittle. Frontal baldness was seen. Senile facial appearance with periorbital puffiness, inverted palpebral fissures and lax skin was noted (Figure 1). Presence of extensive loose folds of skin over face, neck, abdomen, arms and thighs resulting in pendular and sagging skin (Figure 2). The skin of her entire body was flaccid. Stretching of the skin produced delayed recoil. No induration and bruising were noted. There was a left-sided, irreducible, inguinal hernia (Figure 3). Brittle nails were seen over bilateral lower limbs. There was no laxity of joints.

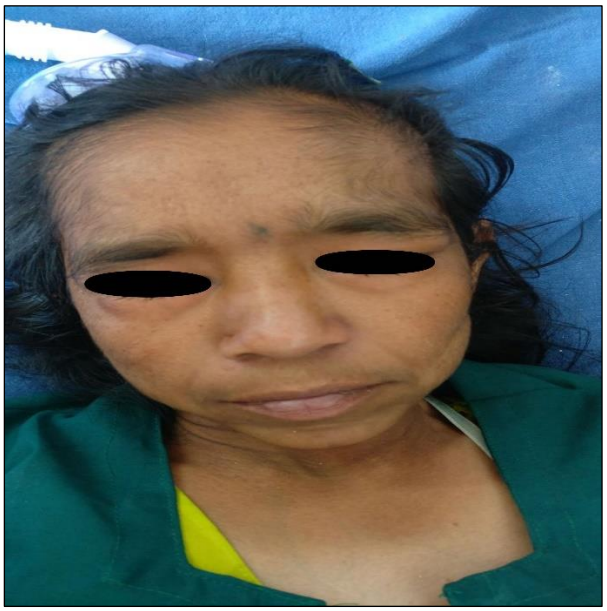

Figure 1: Senile facial appearance with periorbital puffiness, inverted palpebral fissures and lax skin. Frontal alopecia is also noted.

Figure 2: Presence of extensive loose folds of skin over the neck resulting in pendular and sagging skin.

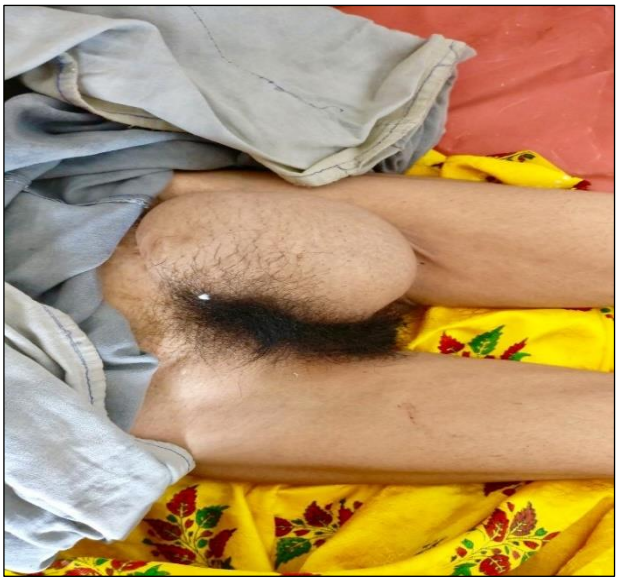

Figure 3: Left-sided inguinal hernia.

Chest examination revealed normal resonance of the lungs bilaterally on percussion and auscultation revealed left infrascapular crepitations. CVS examination: S1 and S2 heard, no murmurs. Per abdomen examination: bowel sounds heard, no organomegaly.

Investigations showed Hb-15.5 g/dl, WBC count-11,830/mm^3, with a DLC of neutrophils-86% and lymphocytes-10%. RBS was 155 mg/dl. Serum electrolytes, RFT and LFT yielded normal results. CRP was 6.41 mg/l. Serological tests for syphilis and HIV were negative. Sputum culture revealed Pseudomonas organisms. Sputum for AFB smear was negative.

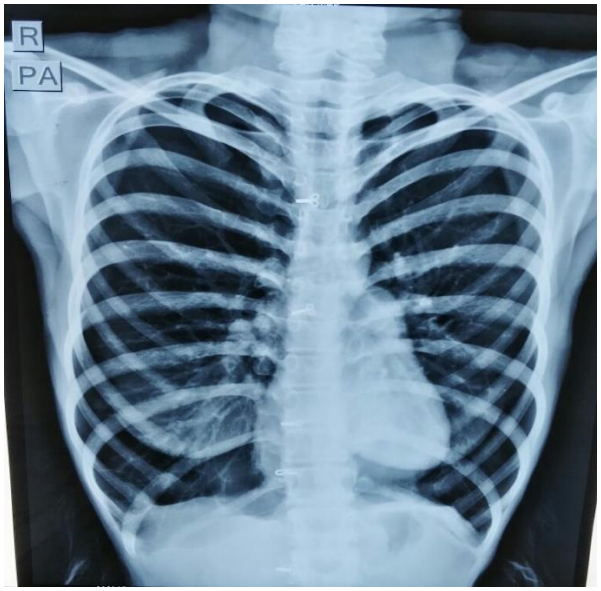

Figure 4: Chest X-ray showing bilateral hyperinflation with widening of intercostal spaces and tubular heart suggesting bilateral emphysematous changes.

A chest x-ray revealed bilateral hyperinflation with widening of intercostal spaces and tubular heart suggesting bilateral emphysematous changes (Figure 4). HRCT showed bilateral bullous changes and retrocardiac left lower lobe cystic changes suggestive of bronchiectasis (Figure 5). Echocardiography showed flail mitral and tricuspid valve with mild mitral regurgitation and mild tricuspid regurgitation, and an ejection fraction of 60%.

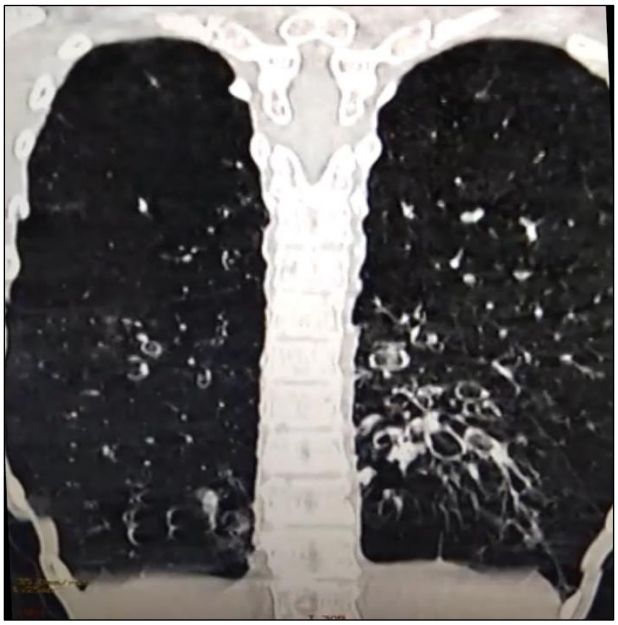

Figure 5: HRCT showing bilateral bullous changes and retrocardiac left lower lobe cystic changes suggestive of bronchiectasis.

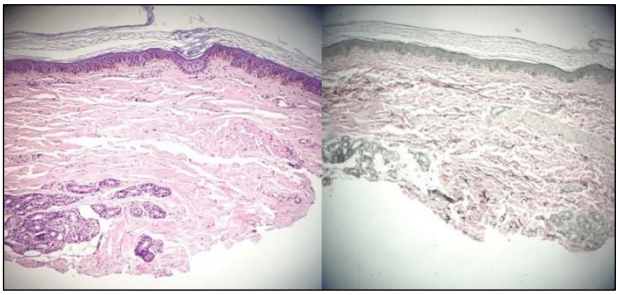

Figure 6: (A) Hematoxylin and eosin stain revealed normal epidermis with fragmented and degenerated elastic fibres in the papillary and reticular dermis and (B) Verhoeff-van Gieson stain showed ratification of elastic fibres.

Biopsy specimens taken from the loose folds of skin over the right arm and left thigh were stained with Hematoxylin and Eosin (H and E) and Verhoeff-van Gieson stain. H and E staining revealed normal epidermis with fragmented and degenerated elastic fibres in the papillary and reticular dermis (Figure 6A). Verhoeff-van Gieson stain showed ratification of elastic fibres (Figure 6B). These findings were consistent with cutis laxa.

The child was treated with IV fluids, Injection Dobutamine, Injection Piperacillin-Tazobactam, Injection Amikacin, bronchodilators, CPAP and emollients.

DISCUSSION

Cutis laxa is rare and occurs due to defects in synthesis of elastic tissue or by its destruction. It can be congenital or acquired.[4] It's typical dermatologic characteristics are loose skin with folds and premature aged appearance. Loss of skin elasticity may involve the entire skin surface. Persons with CL are often described as having a bloodhound-like facial appearance and loose skin of the face, neck, shoulders, and thighs often first attracts attention.[5] A senile aged expression is characteristic and sometimes majority of the body surface is involved.[6]

Congenital cutis laxa can be autosomal dominant, autosomal recessive, or X-linked. Type I autosomal recessive form of congenital cutis laxa shows severe multisystem complications including emphysema, diaphragmatic hernia, gastrointestinal and genitourinary diverticula, and cardiorespiratory complications. Early death can result from cor pulmonale. Hyperextensible skin with slow elastic recoil is the hallmark of cutis laxa. Systemic manifestations are also due to internal elastic tissue abnormalities. Type II autosomal recessive form is a less severe form which is characterized by developmental delay, growth retardation and ligamentous laxity. X-linked recessive form (the occipital horn syndrome) is characterized by mild joint laxity, hernias, bladder diverticula and cranial occipital exostoses.[7] Based on our findings and the history of consanguinity of marriage in the parents, our patient was probably a case of type I autosomal recessive form of congenital cutis laxa.

Differential diagnosis includes acrodermatitis chronica atrophicans, costello syndrome, cutaneous T cell lymphoma, De Barsy syndrome, Ehlers-Danlos syndrome, Focal dermal hypoplasia, Lipodystrophy, Pseudoxanthoma elasticum, Wrinkly skin syndrome, and SCARF syndrome.[8]

Cutis laxa is diagnosed by clinical and histological examination. Histopathological examination shows irregular and fragmented elastic fibers in the reticular layer of the dermis.[4]

CONCLUSION

No medical treatment is available for correction of the pathology of disease or to reduce its progression. Dapsone can be used acutely to control swelling in persons with acquired CL. Plastic surgery remains the only modality of treatment for correction to improve cosmetic appearance and social issues of the patient. Extended facelift, blepharoplasty, and ear lobe reduction may be performed, with obviously, a larger amount of skin excess to remove. Furthermore, these patients should be informed that repeating operations may be necessary.

The systemic abnormalities may need specific treatment depending upon the condition. Botulinum toxin injections are being considered for improving the appearance and facial defects.

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REFERENCES

1. Hajjar WM, Alrajeh AS, Alturki LS, Al-Nassar SA, Hajjar AW. Near-fatal presentation of bilateral pneumothorax in cutis laxa patient: Case report, and review of the literature. Annal Thora Med. 2018;13(4):254.
2. Kariminejad A, Afroozan F, Bozorgmehr B, Ghanadan A, Akbaroghli S, Khorram Khorshid HR, et al. Discriminative features in three autosomal recessive cutis laxa syndromes: cutis laxa IIA, cutis laxa IIB, and geroderma osteoplastica. Inter J Mol Sci. 2017;18(3):635.
3. Irvine AD, Mellerio JE. Syndromes with Premature Ageing. In: Griffiths C, Barker J, Bleiker T, Chalmers R, Creamer D, eds. Rook's Textbook of Dermatology. 9th ed. West Sussex: Wiley-Blackwell; 2016:79
4. do Nascimento GM, Nunes CS, Menegotto PF, Raskin S, de Almeida N. Cutis laxa-Case report. An Bras Dermatol. 2010;85(5):684.
5. Dyer JA. Lipoid proteinosis and Heritable disorders of connective tissue. In: Goldsmith LA, Katz SI, Gilchrest BA, Paller AS, Leffell DJ, Wolff K, eds. Fitzpatrick's Dermatology in General Medicine. 8th ed. New York: McGraw-Hill; 2012:1638-1643.
6. Verhagen AR, Woerdeman MJ. Post-inflammatory elastolysis and cutis laxa. Bri J Dermatol. 1975;92(2):183-90.
7. Choudhary SV, Bisati S, Koley S. Congenital cutis laxa with rectal and uterovaginal prolapse. Ind J Dermatol, Venereol, Leprol. 2011;77(3):321.
8. Dhale SN, Rathod AD, Sonawane S. A case report of cutis laxa. Bombay Hospital J. 2012;54(1):186-7.

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