



Congenital Cutis Laxa with Pulmonary Tuberculosis and Micropenis: A Case Report

Mohammed Akbar Ibrahimi MD¹, khesrow Ekram MD ID^{2,*}, Abdul Ghafar Hamdard MD³, Mohammad Sharif Sediqi MD⁴, Turyalai Hakimi MD ID⁵.

1st Author affiliation, Department of paediatrics, Kabul University of Medical Sciences, Kabul, Afghanistan, mrakbarib1@gmail.com.

2nd Author affiliation, Department of paediatrics, Kabul University of Medical Sciences, Kabul, Afghanistan, khesrowekram@yahoo.com.

3rd Author affiliation, Department of Dermatology, Kabul University of Medical Sciences, Kabul, Afghanistan, ghafarhamdard@hotmail.com.

4th Author affiliation, Department of paediatrics, Kabul University of Medical Sciences, Kabul, Afghanistan, drsharifsediqi@gamil.com.

5th Author affiliation, Department of paediatric surgery, Kabul University of Medical Sciences, Kabul, Afghanistan, Dr.turyalaihakimi@gmail.com.

*Corresponding author: Khesrow Ekram; professor at the Department of paediatrics, Maiwand Teaching Hospital, KUMS, 1st District, Jada-e-maiwand, 1001, Kabul, Afghanistan; Tel: 0093700211454; khesrowekram@yahoo.com. ORCID 0009-0006-4446-3906.

INTRODUCTION

Cutis laxa, which translates from Latin as "loose or lax skin," is an infrequent, hereditary, or acquired condition with no known etiology that is characterized by skin that hangs freely and is completely elasticless [1-2]. Elastolysis in over-all is another name for cutis laxa. Vocal cords, bones, cartilage, blood vessels, the bladder, kidney, digestive system, and lungs are just a few of the organs that are exaggerated systemically. An estimated 1 instance per 2,000,000 people have this extremely infrequent congenital or acquired abnormality [6]. The most obvious sign of aging is loose, pendulous skin, drooping cheeks, or an outward look of immaturity [4-5].

CASE REPORT

A 2-month-old male baby weighing 4.6 kg was admitted to the internal pediatrics ward of maiwand teaching hospital due to sepsis, pulmonary tuberculosis, and cutis laxa. On arrival, he had cough, fever, unable to feed, grunting and cyanosis. On physical examination, there were loose skin, a rectal temperature of 38.8 C, respiratory rate of 85 per minute, a heart rate of 156 per minute, subcostal and intercostal retractions, dyspnea, nasal flaring, bilateral crepitation on the chest, and central cyanosis with oxygen saturation of 84%, cardiovascular examination was normal, and no murmur was heard. Examination of the abdomen was normal with no palpable liver or spleen, further physical examination after admission revealed a loose,

inflexible skin floppy in folds. His face appeared like that of an elderly man (Fig. 1). Loose and baggy skin doubled over the neck, arm, trunk, abdomen, dorsum, and thighs, which gave him a precipitately old man appearance with his skin displaying comprehensible relaxation and elasticity (figs. 2–3–4). The thyroid gland did not look enlarged upon inspection. Patient had a large anterior fontanel, an umbilical hernia, and a micropenis (Fig. 5). On anthropometric measurements, the head circumference was 38 cm, the chest circumference 35 cm, the length 52 cm, and the mid upper arm circumference (MUAC) 11.5 cm. Blood investigations revealed hemoglobin of 10.2 g/dl, a total leucocyte count of 16000/mm³ (polymorphs 85%, lymphocytes 13%, eosinophils 1%, and monocytes 1%), a platelet count of 175000/mm³, and a c-reactive protein of 12 mg/dl. Other laboratory tests revealed normal values for the thyroid function test (Fig. 7), routine urine analysis, blood biochemistry, hepatic enzymes, and serum electrophoresis. Serologic tests for TORCH were negative with abnormal clinical findings; leukocytosis and an elevated level of C-reactive protein were used for the diagnosis of bacterial sepsis and pulmonary tuberculosis. The chest x-ray showed patchy consolidation on both lungs (fig. 6). Further imaging examination revealed normal echocardiography and abdominal ultrasonography. In addition to the aforementioned management, the patient was placed on intravenous normal saline (0.95%) as a maintenance fluid, oxygen, antibiotics consisting of ceftriaxone and ampicillin intravenously, and anti-tuberculosis drugs. After the management of pulmonary tuberculosis and sepsis, he was discharged from the hospital in good condition. Depending on the patient's age and any accompanying issues, the follow-up should be conducted using a

multidisciplinary approach involving pediatricians, dermatologists, plastic surgeons, and pediatric surgeon



Figure 1. A child's face appeared like that of an elderly man.



Figure 2. Shows skin folds in the thighs, legs, and lower parts of the legs in a child with cutis laxa.



Figure 3. Shows the folds in the child's abdomen, which can be easily stretched by the examiner's



Figure 4. Shows excessive stretching of the skin of the upper arm by the hand of the examiner in a child.

Table 1. Shows the normality of the thyroid hormone strain in a laboratory test in a 2-month-old child who is suffering from cutis laxa.



Figure 5. Shows the micropenis and umbilical hernia in a two-month-old baby with cutis laxa.

DISCUSSION

Cutis laxa is a very uncommon malformation that can develop either congenitally or through acquired means [1]. Even though the underlying cause of this aberration is unknown, genetic factors have been linked to its pathophysiology. This condition is known as cutis laxa. Dermomegaly is an abnormality of the elastic fiber network that mostly affects the skin but can also impact other tissues. Loose skin can be seen on the face, dorsum, thighs, neck, hands, and groin with variable degrees of systemic involvement (inguinal hernia, umbilical hernia, micropenis, , large anterior fontanel , and emphysema). Looking at the skin will help you recognize the signs of cutis laxa [7-8].

The patient's family members had a history of a comparable issue (his brother passed away with a related clinical presentation), ruling out autosomal dominant inheritance. His parents were biologically related (second-degree cousins [9–10]). Most likely, type 1 recessive affected our patient. In all the many forms of cutis laxa, he exhibited the recognizable cutaneous anomalies. In addition, he had a micropenis, pulmonary tuberculosis, and an umbilical hernia. Cardiopulmonary abnormalities are frequent in type 1 recessive cutis laxa and are the primary determinants of prognosis and life expectancy. Thankfully, our patient did not have cardiovascular illness, and both the first and second heart sounds during the physical examination and the echocardiography test came back normal. In this instance, our patient had pulmonary TB, which can be attributed to the mother's tuberculosis as a cause. On the other hand, this illness also causes alterations in the child's immune system in addition to skin abnormalities. Given the severity of the patient's condition, first antibacterial therapy did not yield any appreciable results. He was later determined to have TB based on his favorable family history and X-ray results, and a sample of the patient's stomach fluid was aspirated the next



Figure 6. A homogeneous opacity on the right lung and hyperlucency of the left lung are visible on the chest x-ray of a 2-month-old baby.

morning. Gene specialists sent it in for more study, and they discovered the cause.

Fortunately, the patient in our research did not exhibit any indications of diverticula, which are frequently seen in people with Cutis laxa in their urinary and digestive systems.

Endocrine issues and changes in thyroid hormones, which influence children's weight, height, and mental development as well as create constipation and dry skin in addition to causing mental retardation and growth retardation, are linked to Cutis Lexa patients. Thankfully, the patient in question did not exhibit any of the aforementioned symptoms, and the thyroid hormone level was within normal range. Moreover, micropines and an umbilical hernia were observed in this kid, although inguinal hernias were discovered in previous research.

CONCLUSION

This case is significant because of the uncommon documented prevalence of Cutis laxa. The signs of congenital laxity might start to manifest as soon as the kid is born. It has loose skin with creases and a prematurely aged look, which are common dermatologic traits. Skin elasticity loss can affect the entire skin surface, although it is most noticeable on the face, neck, dorsum, and thighs. A systemic symptom, such as pulmonary tuberculosis with micropenis,

ACKNOWLEDGEMENTS

We gratefully acknowledge the staff of the ICU unit of the pediatric internal ward of Maiwand Teaching Hospital, particularly Dr. Mahboob Monshizada,

Dr. Zarin Madani, Dr. Zabihullah Tareen, and Dr. Mostafa Hunaryar, for their cooperation in this study.

CONFLICT OF INTEREST STATEMENT

The authors declare that they have no known financial conflicts of interest or close personal connections that might influence the findings of this study.

FUNDING

The authors received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

ETHICAL APPROVAL

This study was approved by the department of internal pediatrics (protocol number 9, date January 29, 2023), Kabul University of Medical Sciences.

CONSENT

Consent to publish the case report was obtained from the patients' guardians. The Helsinki declaration was taken into consideration.

GUARANTOR

Dr. Khesrow Ekram.

REFERENCES

1. Andiran N, Sarikayalar F, Saraçlar M, Celaglar M. Autosomal recessive form of congenital cutis laxa: more than the clinical appearance. *Pediatr Dermatol*. 2002; 19(4):412-4. <https://pubmed.ncbi.nlm.nih.gov/12383097/>.
2. Balasubrahmanyam G, Yadav J, Ashwini N, Amitha LN, Hanumanthayya K. Acquired cutis laxa with recurrent urticarial. *Journal of Evidence-Based Medicine and Healthcare*. 2015; 2(19): 2977–2981. <https://www.jebmh.com/articles/acquired-cutis-laxa-with-recurrent-urticaria.pdf.pdf>.
3. Chavan SD, Deshmukh AR, Gulanikar AD, Pathrikar SS, Tariq I M. Autosomal recessive cutis laxa type 1 with complex systemic manifestations in Indian. *Journal of Paediatric Dermatology*. 2018; 19(3) 274-276. https://www.researchgate.net/publication/320732424_Autosomal_recessive_cutis_laxa_Type-1_with_complex_systemic_manifestations.
4. Hbib M, Abourazzak S, Idrissi M, Chaouki S, Atmani S, Hida I M. Cutis laxa syndrome: a case report *Pan African Medical Journal*. 2015; 20(3): 1-4. <https://www.panafrican-med-journal.com/content/article/20/3/full>.
5. KunY, Mengdong S, Cong F, Ran H. Congenital cutis laxa: a case report and literature review *Reconstructive and plastic surgery*. 2022; 9: 1-6. <https://pubmed.ncbi.nlm.nih.gov/35372488/>.
6. Ma Y, Zhang J Y, Wang C, Cen W, Liu X, Feng WL. Clinical presentation of a patient with congenital cutis laxa and abnormal thyroid hormone levels. 2014; 6:43–48. <https://www.ncbi.nlm.nih.gov/pmc/articles/pmc3975211/>.
7. Nascimento GMD, Nunes CSA, Menegotto PF, Raskin S, Almeida N. Cutis laxa - case report *An bras dermatol*. 2010; 85(5):684-6. <https://www.scielo.br/j/abd/a/qKX5gWYGMx9sLMYJNgrXZrz/?format=pdf&lang=en>.
8. Rahmati M, Yazdanparast M, Jahanshahi K, Zakeri M. Congenital cutis laxa type 2 associated with recurrent aspiration pneumonia and growth delay: a case report. 2015; 7 (6): 1391–1393. <https://pubmed.ncbi.nlm.nih.gov/26516448/>.
9. Ramu P, Bhavani S R, Kumar D. Congenital cutis laxa with pneumonia: a rare case in pediatric practice. *Journal of the Evolution of Medical and Dental Sciences*. 2015; 4(11): 1889-1892. https://www.jemds.com/data_pdf/1_ramu%205-----suy.pdf.
10. Szabo Z, Crepeau M W, Mitchell A L, Stephen M J, Puntel R A, Yin Loke, et al. Aortic aneurysmal disease and cutis laxa are caused by defects in the elastin gene. *Journal of Medical Genetics*. 2006; 43(3): 255-258. <https://pubmed.ncbi.nlm.nih.gov/16085695/>.