Case Report

Near-fatal presentation of bilateral pneumothorax in cutis laxa patient: Case report, and review of the literature

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Abstract:

Cutis laxa (CL) is a rare connective tissue disease characterized by a loose, wrinkled, and inelastic skin. Here, we report an unusual presentation in a 15-year-old male patient who is a known patient of CL who presented with bilateral pneumothorax. He was successfully managed initially by chest tube insertion and then he was treated surgically with bilateral staged thoracoscopy, apical bullectomy, and pleurodesis with full uneventful recovery.

Keywords:

Bilateral pneumothorax, connective tissue disease, cutis laxa

Cutis laxa (CL) is a rare connective tissue disease characterized by a loose, wrinkled, and inelastic skin. CL could be either congenital or acquired.[1] Congenital type is usually characterized by systemic complications, such as cardiac, pulmonary, and vascular complications, However, an example of these complications are, aortic root dilatation, severe aortic stenosis, or infantile emphysema.[1,2] However, the life expectancy depends usually on the CL type.[3]

Case Report

A 15-year-old male known case of CL, autosomal dominant type based on genetic testing, presented to the emergency department complaining of sudden-onset right-sided chest pain, pleuritic in nature. The pain was associated with dyspnea (Class 2) and dry cough. The clinical examination showed that he has a short stature, as his height was 130 cm and his weight was 38 kg. He has translucent skin with visible veins of the chest and abdomen with no bruising. He has joint laxity of both the upper and lower limbs. There was also a bilateral inguinal hernia [Figure 1].

In the previous surgical history, he had patent ductus arteriosus closure and umbilical hernia repair. His parents were consanguineous (first-degree cousins).

Chest examination showed decrease air entry on the right side and hyperresonance on percussion. However, chest X-ray confirmed large right-sided pneumothorax [Figure 2a], which was treated initially with chest tube insertion, which showed continuous positive air leak. A chest computed tomography (CT) scan confirmed the above diagnosis of large pneumothorax with bilateral apical bullae and saccular ascending aortic aneurysm.

Due to the above findings, he had an emergency operation under general anesthesia, with one-lung ventilation using

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double-lumen tube, right thoracoscopy, apical bullectomy, and pleurodesis. Immediate postoperative recovery was uneventful; however, on the next day postsurgery, he started complaining of sudden onset of severe left-sided pleuritic chest pain and dyspnea. A chest X-ray showed a large left-sided pneumothorax [Figure 2b], which was treated urgently by chest tube insertion. However, the next day, he underwent a similar procedure under general anesthesia and one-lung ventilation, left thoracoscopy, apical bullectomy, and pleurodesis.

Postoperatively, the recovery was uneventful. He was discharged home on day 4 postoperative with a complete expansion of both the lungs [Figure 3].

The histopathology report showed both the right and left lung apices consistent with emphysematous bullae and reactive chronic inflammation.

During his admission, echocardiogram showed moderate aortic root aneurysmal dilation 4 cm in diameter with moderate mitral and aortic valves regurgitation, which was also confirmed by CT scan chest [Figure 4]. Six months later, he was referred to cardiac center and he underwent aortic root surgery due to aneurysmal aortic root dilatation and regurgitation.

Discussion

CL is a rare connective tissue disease characterized by a loose, wrinkled, and inelastic skin. CL could be either congenital or acquired.[1] The congenital type of CL includes autosomal dominant, autosomal recessive Type 1, and autosomal recessive Type 2.[2] CL autosomal dominant was characterized by cardiac and pulmonary complication such as emphysema and aortic root dilatation, and usually, they have a normal life expectancy.[3] CL autosomal recessive Type 1 was associated with severe systemic complication, which includes severe aortic stenosis and infantile emphysema that has a poor prognosis.[4] CL autosomal recessive Type 2 was characterized by abnormal elastic skin, joint deformity, and delay development.[5] The acquired form of CL was characterized by premature aging with or without systemic involvement.[6] Nascimento *et al*. reported a case of a hereditary form of CL in a 33-month-old girl with unilateral pneumothorax and left inguinal hernia.[4] However, Genevieve *et al*. reported different presentations of fatal CL presented with severe emphysema complicated by lung infection, leading to patient death at the age of 10 months.[7]

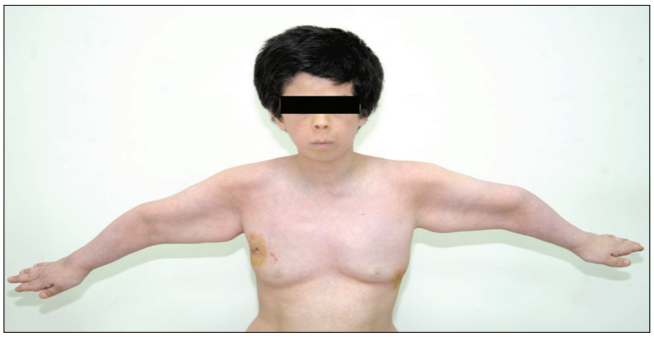

Figure 1: Photograph of the patient showing translucent skin with visible veins in the chest and abdomen, joint laxity of the upper and lower limbs

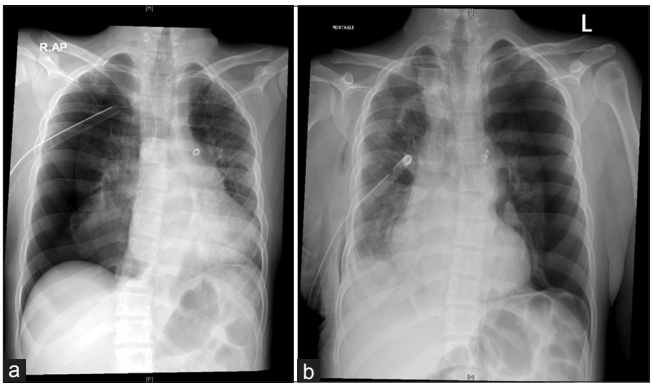

Figure 2: (a) Chest X-ray showed a large right-sided pneumothorax with chest tube. (b) Chest X-ray 1-day postoperative showed the large left-sided pneumothorax

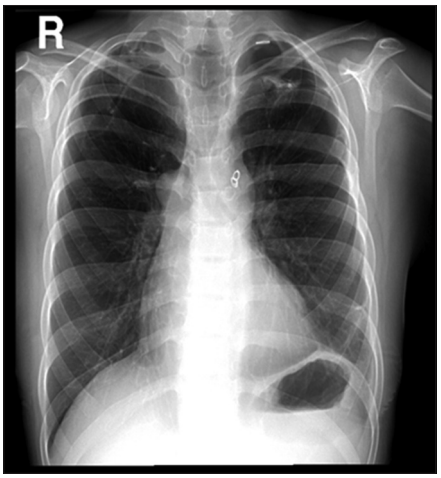

Figure 3: Chest X-ray postoperative showed complete expansion of both the lungs

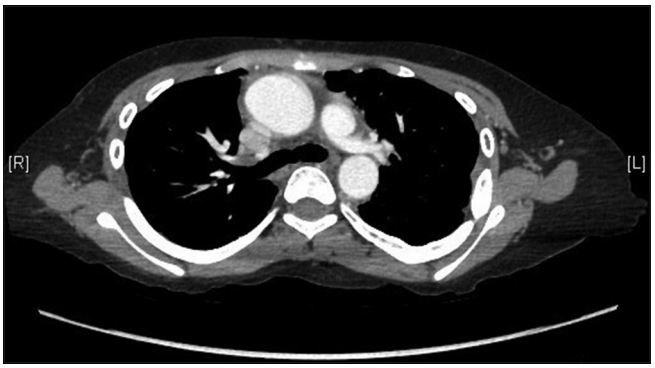

Figure 4: Computed tomography scan of the chest showed moderate aortic root aneurysmal dilation 4 cm in diameter

Other presentation of CL autosomal recessive Type 1 reported by Hbibi *et al*. of 18-month-old boy with aortic dilation, right side inguinal hernia, scoliosis, and emphysematous lungs.[8]

Our patient was a 15-year-old boy known with a CL autosomal dominant type; he has a very rare form of near-fatal presentation with acute bilateral pneumothorax, which was treated successfully initially by chest tube insertion and then by bilateral staged thoracoscopic surgery, apical bullectomy, and pleurodesis. He made full recovery after surgery without any complications, and he remained very well 2 years on regular follow-up in the outpatient clinic. To the best of our knowledge, this is the first time reported in the literature of this kind of acute presentation in a CL patient.

Ethics or IRB

A valid written consent was obtained from his guardian.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Acknowledgment

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Nil.

Conflicts of interest

There are no conflicts of interest.

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