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CASE REPORT Open Access

Congenital absence of sternum with tetralogy of Fallot, right aortic arch, and bilateral superior vena cava

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Abstract

Background Congenital complete absence of sternal bone or cleft sternum or absent sternum is a very rare and potentially life-threatening birth defect. An association of cleft sternum with tetralogy of Fallot, right aortic arch, and bilateral superior vena cava is extremely rare, and not many cases have been reported.

Case presentation A 2-year-old child was brought for routine checkup. He was a known case of congenital heart disease and midline thoracic defect at age of 3 months. Further investigations revealed absent sternum with tetralogy of Fallot, large cono-ventricular defect with bidirectional shunt, severe infundibular pulmonary stenosis, superiorly located right aortic arch, and bilateral superior vena cava.

Conclusion Congenital absence of the sternum with intracardiac defects like tetralogy of Fallot with right aortic arch and bilateral superior vena cava is extremely rare malformation. Early diagnosis and surgical correction give the infant the best chance of survival.

Keywords Cleft sternum, Congenital heart disease, Midline defect

Background

Complete congenital sternal cleft or cleft sternum (CS), also called absent sternum, is a very rare and potentially life-threatening birth defect occurring due to failure of fusion of sternal bone [1]. It can be partial or complete and can be isolated or associated with cardiac and noncardiac anomaly like defect of abdominal wall, diaphragm, pericardium, and the heart [2]. An association of complete CS with tetralogy of Fallot (TOF), right aortic arch, and bilateral superior vena cava (SVC) is extremely rare, and not many cases have been reported. Early diagnosis and surgical correction give the infant the best chance of survival.

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Case presentation

A 2-year-old child was brought to outpatient services by his father for routine checkup. He was a known case of congenital heart disease and midline thoracic defect at age of 3 months.

There was a prominent midline raphe over the skin starting from anterior neck and visible cardiac pulsations in the suprasternal region (Figs. 1 and 2). Examination revealed underweight child with a central bony defect under the depression along the midline and a grade II systolic murmur over the precordium. The rest of the examination was normal with no other obvious congenital anomalies. The baby was hemodynamically stable. This led to the diagnosis of congenital absence of the sternum.

Chest radiograph (Fig. 3) revealed mild cardiomegaly. 2D echo revealed TOF, large cono-ventricular defect with bidirectional shunt, severe infundibular pulmonary stenosis, superiorly located right aortic arch, and bilateral SVC. Left SVC draining to dilated coronary sinus.

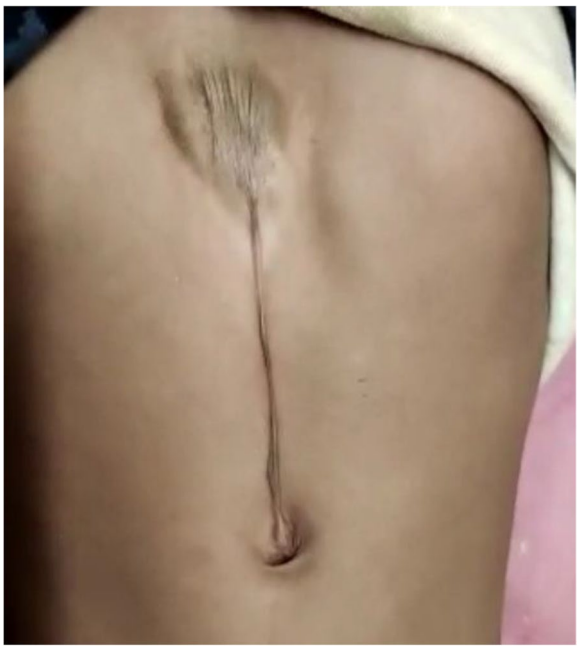

Fig. 1 The absent sternum along with visible cardiac contractions

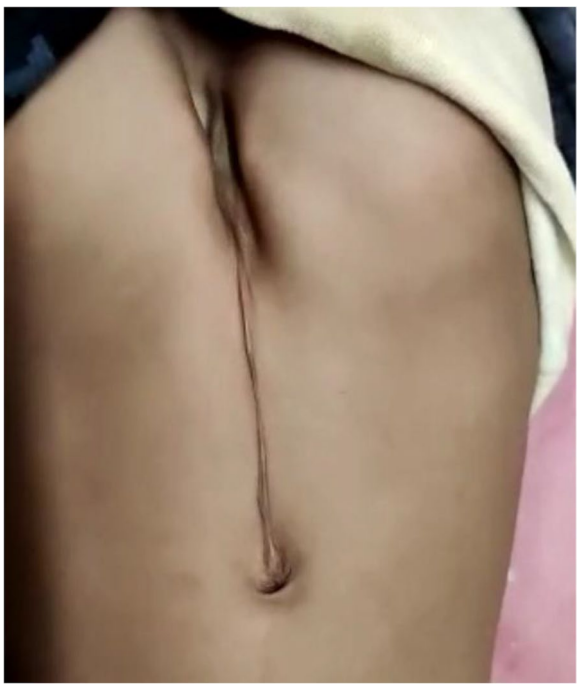

Fig. 2 Vasculature was seen beneath a thin layer of the skin

A computed tomography (CT) scan of the thorax (Fig. 4) was done to rule out any other associated malformations. The scan confirmed a complete CS in anterior chest wall with defect of 4.5 cm through which

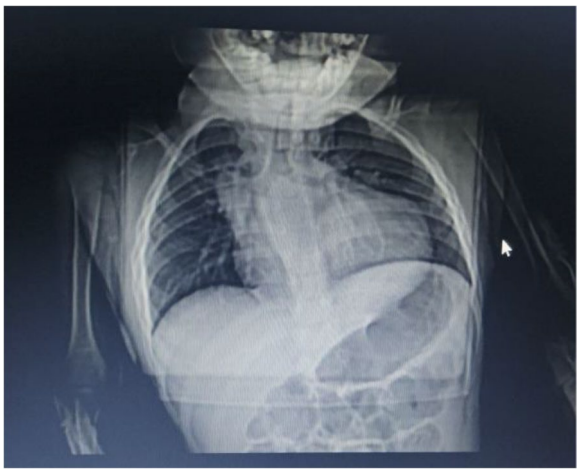

Fig. 3 Chest radiograph

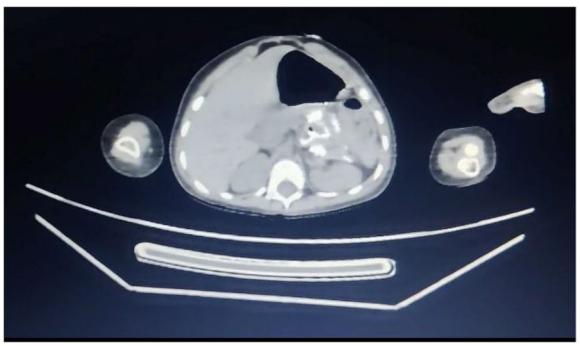

Fig. 4 CT scan chest

anterosuperior portion of great vessel and anterior portion of heart seen herniating and no other congenital malformation. CT scan of the brain was normal. Sonography of abdomen and pelvis was normal.

Cardiac surgery opinion advised extremely high-risk intracardiac repair with sternal closure. Parents refused for further surgical management due to extremely high-risk prognosis.

Discussion

Complete congenital sternal cleft (CS), also called absent sternum, is a very rare and potentially life-threatening birth defect [1]. It results from failure of the process of midline mesenchymal strip fusion during embryonic development [2]. It accounts for only 0.15% of all chest wall malformations. It can be complete and incomplete type.

Complete CS is very rare and involves splitting of the sternum from the manubrium to the xiphoid. This variety is also strongly associated with complex intracardiac defects such as TOF, Cantrell's pentalogy, ectopia cordis,

and many others. Noncardiac anomalies may be cervicofacial hemangiomas, diastasis recti, and cleft mandible. Incomplete CS presents with chondral bridges either in the upper part (inferior-type incomplete CS) or in the lower part (superior-type incomplete CS) of the sternum. It is very often associated with vascular dysplasias. A supraumbilical raphe is present in 30% of cases of CS. The etiology of CS is speculative.

There are two theories involved:

- Failure of fusion of lateral sternal bands with midline mesodermal structures between the 6th and 9th gestational weeks.
- Early amnion rupture may result in multiple defects by pressure necrosis, incomplete embryogenesis, and tearing and tethering by amnion bands. Surgery is advocated to protect the underlying heart from external trauma to allay parental anxiety and to prevent recurrent respiratory tract infections [3].

CS might occur in isolation or along with defects of abdominal wall, diaphragm, pericardium, and heart. These patients are at increased risk of mediastinal trauma, hypothermia, increased insensible fluid losses, cyanosis, and recurrent infections of the chest [2].

Published experience with absent sternum is limited to sparse case reports, and no preferred method for management has been described [1].

Early diagnosis and surgical correction give the infant the best chance of survival [4]. Isolated CS is best repaired primarily in the neonatal period because extreme compliance and pliability of bony thorax permit direct sternal closure. However, the presence of complex cardiac defects postpones the sternal repair until the age and weight of elective operation of the cardiac malformation are reached [3, 5]. Reconstructive surgery of absent sternum should be performed by primary closure using combined periosteal advancement flap and sliding osteochondroplasty during the neonatal period when the chest wall is highly compliant and closure can be achieved without significant cardiopulmonary compromise [2].

In our case, combination of complete sternal cleft associated with intracardiac defects like tetralogy of Fallot with vascular anomaly like right aortic arch and bilateral vena cava obviously makes it extremely rare.

Conclusion

Congenital absence of the sternum with intracardiac defects like TOF with right aortic arch and bilateral vena cava is extremely rare malformation. Early diagnosis and surgical correction give the infant the best chance of survival.

Abbreviations

CS Cleft sternum
SVC Superior vena cava
TOF Tetralogy of Fallot

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Authors' contributions

SC has contribution in collecting the patient data and writing manuscript. NN has analyzed and interpreted data and also has contribution in writing manuscript. RW has analyzed data and also has contribution in writing manuscript. All authors read and approved the manuscript.

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Availability of data and materials

Available (all the investigations and reports of the patient are available). All the data and materials-available with the hospital record-keeping section written on indoor papers. Also, it is available with the patient.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Written informed consent taken.

Competing interests

The authors declare that they have no competing interests.

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