

Congenital absence of the sternum in a neonate

Nirmala Cherukuri¹, Mallesh Gampa², Swapna Lingaldinna³, Himabindu Singh⁴

From ¹Professor, ²Junior Resident, Department of Paediatrics, ³Assistant Professor, ⁴Professor, Department of Neonatology, Niloufer Hospital, Hyderabad, Telangana, India

Correspondence to: Dr. Swapna Lingaldinna, Department of Neonatology, Niloufer Hospital, Lakdikapool, Redhills, Hyderabad, Telangana, India. E-mail: drswapnalingaldinna@gmail.com

Received - 18 May 2019

Initial Review - 10 June 2019

Accepted - 17 June 2019

ABSTRACT

Congenital absence of the sternum is a rare chest wall malformation resulting from the failure of midline fusion during embryonic development. It is a potentially life-threatening congenital midline defect. Only sporadic cases have been reported in literature. The abnormality can cause significant morbidity, and like other congenital anomalies can have associated defects. Repair of congenital absence of the sternum should ideally be undertaken in the neonatal period when the chest wall is highly compliant, and hence, primary closure can thus be achieved without significant cardiopulmonary compression. As the patient ages, chest wall compliance decreases and closure will become progressively difficult as venous return and lung compliance are increasingly compromised. We report a case of congenital absence of the sternum as it is very rare and because it was successfully operated in a neonate period.

Key words: Anomalies, Cantrell's pentalogy, Congenital absence of sternum, Neonate

Congenital absence of the sternum is a potentially life-threatening congenital midline defect resulting from the failure of midline fusion during embryonic development [1]. The earliest embryologic evidence of the sternum can be found at 6 weeks of gestation; at this time, it appears as two parallel lateral mesenchymal bands that arise in the anterior thoracic wall independently. By the 10th intrauterine week, the sternal bands fuse craniocaudally in the midline to form the body of the sternum and part of the manubrium [2-5]. The absence of the sternum can result in triggering of multiple risks, the most common being mediastinal injury, hypothermia, and insensible fluid losses. Equally severe risks factors that can be driven by the absence of the sternum are respiratory and hemodynamic sequelae.

Isolated congenital absence of the sternum is an extremely rare condition. Its estimated incidence is unknown; although, it is rarer than the Cantrell's pentalogy. Other associated defects of midline fusion are also included in the various anomalies interlinked with sternum clefts, the most significant of which is Cantrell's pentalogy, also known as thoracoabdominal syndrome, is a rare condition that drives defects involving the diaphragm, abdominal wall, pericardium, heart, and lower sternum. Possibility of the occurrence of the certain unrelated process such as the PHACES syndrome, consisting of posterior fossa malformations, arterial lesions, cardiac, and eye disease is also there. The incidence rate of Cantrell's pentalogy has been estimated to be around 1 in 100,000 live births and which includes defect of sternum, diaphragm, pericardium, and intracardiac defects. Here, we report a case of congenital absence of the sternum in a neonate.

CASE REPORT

A term neonate born at 38 weeks of gestation with a birth weight of 3.2 kg was referred at the 2nd h of life with anterior chest wall deformity. The neonate born out of normal vaginal delivery was born to a primi mother with no significant family history and with an uneventful antenatal period with no detection of any abnormalities in the antenatal scans.

On examination, the neonate had a defect in the sternum of 4 cm in width which was covered by a yellowish membrane. The associated paradoxical movement was observed along with the visible cardiac contractions beneath a thin layer of the membrane (Fig. 1). The abdomen and other systemic examinations were normal along with ultrasound abdomen and two-dimensional echo, and no other internal or external congenital abnormalities were present. The absence of the sternum was observed through chest computed tomography scan where other thoracic structures were normal. After initial stabilization, the neonate was operated by the team of cardiothoracic and plastic surgeons. The neonate underwent chest wall reconstruction with bilateral pectoralis major muscle flaps. A skin incision was given around the scab, and the flaps were created. Pectoralis major flaps were created bilaterally, and the scab was excised to further approximate the edges of the defect. Pectoralis major flaps were approximated in the midline in a double-breasted fashion followed by closing the skin incision. The post-operative period was uneventful. The neonate was followed regularly and now is 1 year old, with normal development and stable anterior chest (Fig. 2).



Figure 1: Neonate with a defect in the sternum



Figure 2: Follow-up of the neonate after 1 year

DISCUSSION

Congenital absence of sternum/complete sternal cleft, is a rare chest wall malformation, with the incidence of <1% of all chest wall malformations [1]. At 5–6 months of embryonic development, the sternum begins to chondrify and then subsequently ossifies. This ossification begins as a single center at the manubrium and proceeds in a cephalocaudal direction. In the body of the sternum, the ossification centers are usually observed in pairs, and this process is usually completed within the 1st year after birth [6]. Although the embryologic development of the sternum has been studied in detail, the etiology of the cleft sternum is unknown and has not yet been established. Some hypotheses include chronic nutritional deficiency, a lack of riboflavin during pregnancy, and disruption of the *HoxB4* gene have been reported [7]. Most clefts occur sporadically, although, in 1984, Haque reported an autosomal recessive familial association.

The absence of the sternum is associated with defects which include maxillofacial hemangiomas, cleft lip or palate, pectus excavatum, precordial skin tags, gastroschisis, and other defects such as coarctation of the aorta, cardiac defects, eye abnormalities, posterior fossa anomalies, and hidden hemangiomas [8]. In some cases, it is a part of PHACES syndrome or Cantrell's pentalogy [9].

The pentalogy of Cantrell consists of a midline, supraumbilical abdominal wall defect, a defect of the lower sternum, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm, and intracardiac anomalies.

Clinically, infants with a sternal cleft have a concave midline thoracic defect, which can be associated with the paradoxical movement of the chest wall during respiration. In the neonatal period, neonates are mostly asymptomatic. On physical examination, a paradoxical midline thoracic bulging (protrusion of the mediastinal viscera during expiration) is noticed. The changes in intrathoracic pressure that result from this paradoxical movement can cause displacement of the heart and large vessels and impairment of venous return. As a result, the infants may have symptoms of right ventricular overload, cyanosis, respiratory distress, and arrhythmias. Furthermore, the infants have reduced aeration of the lungs and decreased the force of the cough reflex, which may lead to recurrent upper respiratory tract infections. In addition, cardiac pulsations can sometimes be seen through the thin and ulcerated skin that overlies the defect.

The thoracic wall has higher compliance in neonates, making a primary closure easier without any other additional procedures such as chondrotomies, osteotomies, or clavicular dislocation. Even the risk of cardiovascular impairment is lower, the baby was operated with an uneventful post-operative period, which further re-establishes the positive outcomes observed from surgeries generally observed in the neonatal period. The outcome of the present case was similar to the cases reported by Kanojia *et al.*, where two babies were reported to be operated in the neonatal period with a good post-operative period [10].

In the present case, the diagnosis was not picked up in the antenatal period, whereas in a case reported by Héron *et al.*, the patient was diagnosed with sternal cleft by ultrasound at 21 weeks' gestation [11]. It appears that screening of the abnormalities is more accessible when there is an association of cardiac anomaly. In a case report by Harjai, there was a necessity of partial removal of the thymus for space constraints in a newborn, but in the present case, the surgery consisted only in the chest wall closure, and it was well-tolerated [12]. In elderly patients, the rib cage is stiff and repair is difficult. Increase in thoracic wall compliance to allow primary closure can be attained through sliding chondrotomies, osteotomies, or clavicular dislocation [13,14].

CONCLUSION

Congenital absence of the sternum is a rare malformation of the chest wall which results from failure of the process of midline mesenchymal strip fusion during embryonic development. Patients with a complete sternal cleft are at increased risk of mediastinal trauma, hypothermia, increased insensible fluid losses, cyanosis, and recurrent infections of the chest. With increasing age, surgical correction becomes difficult due to a decrease in compliance of the chest wall necessitating early surgical correction in the neonatal period.

REFERENCES

1. Eijgelaar A, Bijtel JH. Congenital cleft sternum. *Thorax* 1970;25:490-8.
2. Klima M. Early development of the human sternum and the problem of

- homologization of the so-called suprasternal structures. *Acta Anat (Basel)* 1968;69:473-84.
3. Haque KN. Congenital absence of the sternum. *Arch Dis Child* 1979;54:905-6.
 4. Torre M, Rapuzzi G, Carlucci M, Pio L, Jasonni V. Phenotypic spectrum and management of sternal cleft: Literature review and presentation of a new series. *Eur J Cardiothorac Surg* 2012;41:4-9.
 5. Hu Y, Wang Y, Luo Z, Li C. A new symmetrodont mammal from China and its implications for mammalian evolution. *Nature* 1997;390:137-42.
 6. Haque KN, Zaidi MH. Congenital absence of sternum in a newborn infant, and its possible aetiology. *Trans R Soc Trop Med Hyg* 1980;74:266.
 7. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. *Surg Gynecol Obstet* 1958;107:602-14.
 8. Mазzie JP, Lepore J, Price AP, Driscoll W, Bohrer S, Perlmutter S, *et al.* Superior sternal cleft associated with PHACES syndrome: Postnatal sonographic findings. *J Ultrasound Med* 2003;22:315-9.
 9. Sarper A, Oz N, Arslan G, Demircan A. Complete congenital sternal cleft associated with pectus excavatum. *Tex Heart Inst J* 2002;29:206-9.
 10. Kanojia RP, Wakhlu A, Wakhlu A. Isolated cleft sternum: Neonatal surgical treatment. *J Indian Assos Paediatr Surg* 2007;12:89-91.
 11. Héron D, Lyonnet S, Iserin L, Munnich A, Padovani JP. Sternal cleft: Case report and review of a series of nine patients. *Am J Med Genet* 1995;59:154-6.
 12. Harjai MM, *et al.* Management of space constraints in sternal cleft repair. *J Pediatr Surg Space* 2008;2:28-30.
 13. Zamfir C. Sternal cleft – A rare congenital malformation. *J Paediatr Surg Case Rep* 2014;2:97-100.
 14. Samarrai AA, Charmockly HA, Attra AA. Complete cleft sternum: Classification and surgical repair. *Int Surg* 1985;70:71-3.

Funding: None; Conflict of Interest: None Stated.

How to cite this article: Cherukuri N, Gampa M, Lingaldinna S, Singh H. Congenital absence of the sternum in a neonate. *Indian J Child Health.* 2019; 6(7):394-396.

Doi: 10.32677/IJCH.2019.v06.i07.016