CASE REPORT

The demonstration of the inferior sternal cleft using three-dimensional reconstruction: a case report

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Abstract

Congenital sternal cleft is a rare disorder in which there is a gap in the midline of the anterior chest wall between the two halves of the sternum. Typically, the contour of the mediastinal structures can be seen beneath the skin. It is rare and the exact incidence is not known. It results from failure of fusion of the two lateral mesodermal sternal bars by 8 weeks of gestation. Most cases are diagnosed shortly after birth and are reported only rarely in adults. We report here one of the congenital major chest wall deformities; inferior sternal cleft is rarely seen, associated with sternal and costal variations in a 22-year young man.

Keywords: sternum, chest wall deformity, cleft sternum, sternebrae, skeletal variation.

Introduction

Sternal malformations are rare congenital malformations of the chest wall. Only a few cases have been reported. Most cases are diagnosed shortly after birth and are reported only rarely in adults. Ravitch MM *et al.* [1, 2] reported 47 cases found in the literature between 1888 and 1977. In one series of 5182 chest wall defects, 0.15% were sternal clefts. Seventy-five percent of these patients were female [3]. Isolated sternal cleft is a separate entity from ectopia cordis and other midline fusion deformities. It may be seen in association with craniofacial hemangiomas or rarely with other vascular malformations, such as aneurysm of the ascending aorta or obliteration of the right innominate artery [4, 5]. There is no known causal gene, teratogen, or nutritional deficiency. However, a mouse model in which the Hoxb-4 gene was mutated yielded cleft sternum in the mouse [6].

Sternal clefts may be classified as follows: (1) complete cleft sternum without additional malformations or with ectopia cordis, large vessel, facial, and cerebrovascular anomalies; (2) superior sternal clefts without additional malformations or with anomalies as in cases of total clefts; (3) inferior sternal clefts with additional severe malformations (Cantrell's syndrome [7] or (4) sternal clefts within the scope of a total ventral cleft.

Inferior sternal cleft usually occurs in Cantrell's pentalogy, which also includes an omphalocele-like abdominal defect, a crescent-shaped anterior diaphragmatic defect, and a hole in the pericardium, which allows pericardial-peritoneal communication. Cantrell's pentalogy is also often associated with various intra-cardiac anomalies, such as atrial or ventricular septal defect, tetralogy of Fallot, and ventricular diverticulum [2, 8].

We report a rare case of inferior sternal cleft and absent processus xiphoideus in an adult male not associated with cardiac defects or ectopia cordis. Furthermore, we detected that sternum was more inferior than usual skeletal level (T5–T9 vertebrae); important costal variations were also reported with regard to its costosternal joints.

Patient, Methods and Results

In this study, we reported inferior sternal cleft, rare congenital chest wall deformity caused by failure of fusion of sternal elements, and absent processus xiphoideus in a 22-year young man who was referred to Department of Neurology with his head and neck pain complaints, including obtaining a written consent from the patient. Because our patient applied into a hospital for his complaints fort he first time, we have not known the patient's history. We know verbally that he had not any complaints at birth, during childhood and adolescence. Our patient has no brother or sister. On physical examination, his mother and father have no skeletal defects in thorax.

It was determined that bone structures of his chest, especially sternum and ribs, seemed more lower than its normal anatomic skeletal levels on anterior chest wall. In addition, clavicle was also more oblique and longer

(15.17 cm in the right side; 15.47 cm in the left side) than its normal anatomic position. First of all, it was thought that patient had a congenital chest wall deformity, namely pectus excavatum, is the most prevalent of the anterior chest wall deformities and the most extensively studied and reported. It has also been called "funnel chest" or "trichterbrust". Routine medical examinations were performed respectively. The values of pulmonary function tests (resting spirometry), exercise testing, and echocardiography were within normal clinical limits. In computed tomography findings, the sternum was located between at the upper border of T5 and at the lower border of T9-vertebrae. Normally, the upper border of the manubrium is marked by the incisura jugularis, which is easily palpable and is usually at the level of the T3-vertebrae. Below, the manubrium sterni articulates with corpus sterni at the angulus sterni, which is marked by a palpable (and sometimes visible) transverse ridge about 5 cm below the incisura jugularis. In our case report, this transvers ridge was clearly visible and 5.8 cm below the incisura jugularis (Figure 1).

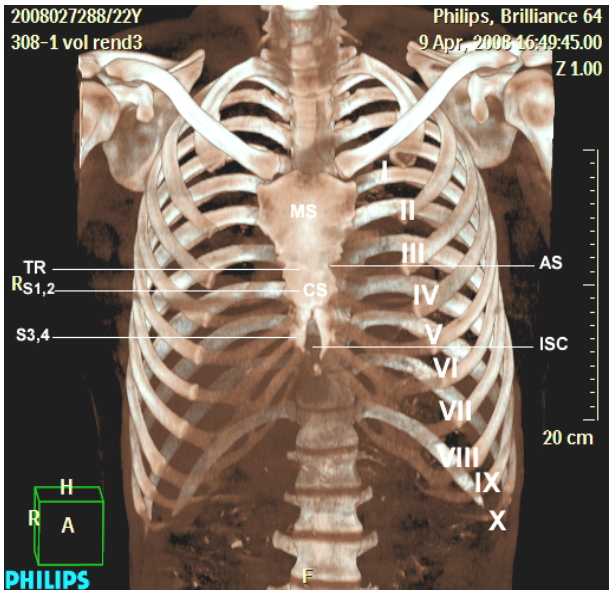

Figure 1 – ***Computed tomographic chest roentgenogram with three-dimensional reconstruction that revealed the corpus sterni had two sternebrae instead of the usual four (MS: manubrium sterni; CS: corpus sterni; TR: transvers ridge at the level of symphsis manubriosternalis; AS: angulus sterni; ISC: inferior sternal cleft; S1,2: first and second sternebrae; S3,4: third and forty sternebrae; I–X: costae I–X).***

This length also indicated the length of manubrium sterni. Furthermore, the width of manubrium sterni (7.5 cm) looked wider than usual at the level of the first costa cartilagine (Figure 1). The angulus sterni is an important bony landmark at the T4- or T5-vertebral level. It indicates not only the manubriosternal junction but also the level of the second costal cartilages; hence, it is a reference point in counting ribs. Rarely, however, the angulus sterni is at the level of the third costal cartilages. We also determined that the angulus sterni received the third costa cartilagine at the lower border of T7-vertebra (Figures 1 and 2). The manubrium sterni received first and second costa cartilagine (Figures 1 and 2). Anatomically, it should have received only first costa cartilagine.

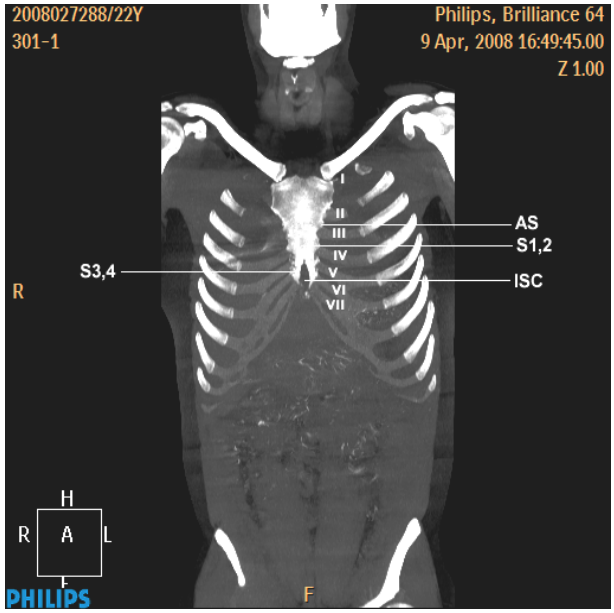

Figure 2 – ***Computed tomographic chest roentgenogram with three-dimensional reconstruction that revealed costal variation clearly. The manubrium sterni and corpus sterni received I.–II. costal cartilages and IV.–VII. respectively. And, the angulus sterni received III. costa cartilagine instead of II. costa cartilagine (AS: angulus sterni; S1,2: first and second sternebrae; S3,4: third and forty sternebrae; ISC: inferior sternal cleft; I–VII: costae I–VII).***

In normal adult, the corpus sterni, about twice as long as the manubrium sterni, is notched on each side to receive costal cartilages 2 to 7. In our case, it appeared that only two sternebrae exist instead of the usual four. The corpus sterni received only 4.–7. costal cartilages (Figures 1 and 2). Anatomically, it should have received only 3.–7. costa cartilaginea. The sternum originally consists of two cartilaginous bars, situated one on either side of the median plane and connected with the cartilages of the upper nine ribs of its own side. These two bars fuse with each other along the middle line to form the cartilaginous sternum, which is ossified from six centers: one for the manubrium, four for the body, and one for the xiphoid process. One of the findings in our patient was that the lower portions of the original cartilaginous bars failed to fuse leaving the wide triangular gap in the midline. Thus, what appear to be "the processus xiphoideus" in this patient was actually unfused 3rd and 4th sternebrae, and the processus xiphoideus had never developed. Because, the processus xiphoideus are usually not that big and they do not normally receive the ribs. More common defect was inferior sternal cleft where the defect was seen in the midline, and not a triangular gap (Figures 1–3). The other finding was that 3rd and 4th ossification centers were missing, developmentally making the corpus sterni very short (Figures 1 and 2). The length of corpus sterni through the beginning of inferior sternal cleft and the length of inferior sternal cleft was 3.96 cm and 2.12 cm respectively. Totally, the length of the sternum was 11.88 cm.

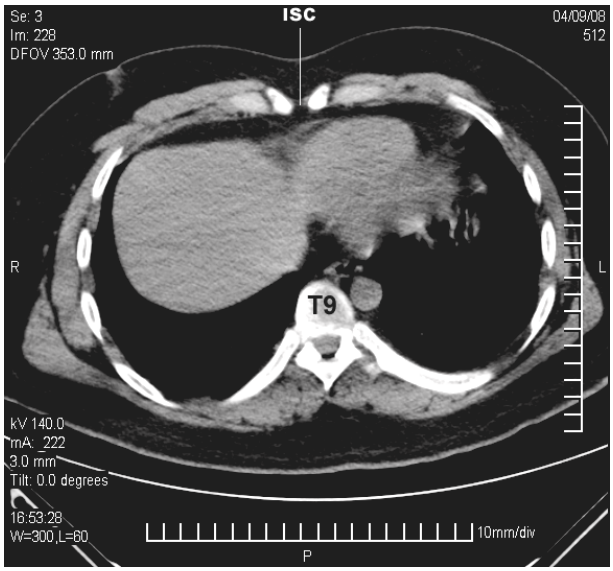

Figure 3 – ***Chest CT of patient with inferior sternal cleft at the level of T9 vertebra.***

Discussion

The sternum is an elongated, flattened bone, forming the middle portion of the anterior wall of the thorax. Its upper end supports the clavicles, and its margins articulate with the cartilages of the first seven pairs of ribs. It consists of three parts, named from above downward, the manubrium sterni, the corpus sterni or gladiolus, and the processus xiphoideus; in early life, the corpus sterni consists of four segments or sternebrae. Its average length in the adult is about 17 cm, and is rather greater in the male than in the female.

During embryonic development, the sternum arises independently of the ribs from paired mesenchymal bar presents by 6 weeks gestation. These parallel bars migrate to midline, where they undergo chondrification and fusion by 9 weeks gestation. The fusion process occurs in cephalo-caudal direction and is followed by approximation of the ventrally growing ribs. Independent of the development of the sternal bands, a single midline condensation of mesenchyme develops, which later forms the manubrium sterni. Sternal defects result from failure of fusion of the sternal bands. This process appears to be distinct from that, leading to cephalic body wall closure. The etiology of inferior sternal clefts associated with other defects of the cephalic fold and abnormal development of the septum transversum, such as Cantrell's pentalogy, is not well understood. It is clear that inferior sternal clefts are embryologically different from superior and complete sternal clefts [9, 10].

Sternal clefts are extremely rare, and because their exact incidence is unknown. In the largest series of 47 cases reported by Ravitch MM [1, 2], 16 were superior and 31 were complete. Cases have been reported from all parts of the world, with a seemingly high frequency of reports from the Middle East similarly as in our report [11]. The etiology of the sternal clefts is not at all clear. The majority of cases are sporadic. Haque KN noted nutritional deficiency in the mothers of several of his patients and postulated that riboflavin deficiency might have been responsible for the skeletal defects [11]. However, it is of interest that two of his families were consanguineous, and in one, there was a recurrence, suggesting that rarely this condition is inherited in an autosomal recessive manner. Gorlin RJ *et al.* reported sisters, both with nonunion of the sternum, with a teardrop shaped umblicus in the first and hemangiomas in the second [12]. In the same study, Gorlin RJ *et al.* reviewed the association of the sternal clefts with supraumblical raphe and found 42 examples in the literature from 1842 to 1992. No sex predilection was noted in this group, which did not have facial hemangiomas. A second group of 31 patients reported between 1880 and 1994 had a marked female predominance (29 women; two men) and had facial hemangiomas appearing within the first weeks of life, an associated first recognized by Hersh JH *et al.* [4]. Within this second group is an apparent syndrome, recently termed PHACE syndrome, which includes the association of posterior fossa brain abnormalities (typically Dandy-Walker malformation), hemangiomas, arterial malformations, coarctation of the aorta, cardiac malformations, and eye malformations [13, 14].

Currently, the etiology of sternal cleft remains, in most cases, unknown. The high frequency of cases from the Middle East and the report of consanguinity and recurrence in at least one family suggest that an autosomal recessive gene is rarely responsible for this defect, and this issue should be considered in genetic counseling. Since the thoracic viscera are covered, detection by elevations in maternal serum alpha-fetoprotein would not be likely. Ultrasound prenatal diagnosis of sternal abnormalities associated with complete ectopia cordis and inferior clefts associated with Cantrell's pentalogy of course have been reported, but this is apparently of a different embryologic origin.

Partial or complete sternal cleft is a rare congenital anomaly. The development of the rest of the sternum and its association with the ribs laterally remains normal. On the contrary, we reported important sternal and costal variations associated with inferior sternal cleft. We noted that sternum was inferior localization between at the upper border of T5 and at the lower border of T9-vertebrae and the processus, xiphoideus never had developed. The other important point was the level of the angulus sterni receiving the third costa cartilagine at the lower border of T7-vertebra.

A cleft sternum may be partial or total. Three types of sternal cleft may be noted on examination. Superior cleft sternum is most common. It is an incomplete defect that involves the upper sternum or manubrium. The inferior aspect of the sternum is fused, which creates a U- or V-type deformity. In the V-deformity, there is often only a narrow bridge at the xiphoid process, which is sometimes referred to as a subtotal sternal cleft. Patients who have this abnormality also may have a midline raphe or band-like scar that extends to the umbilicus [3]. Usually, it is not associated with cardiac defects and is a relatively benign condition. Rarely is there an isolated fissure in the caudal portion of the sternum without other congenital anomalies. This anomaly is ascribed to premature termination of an

otherwise normal course of development. Central perforations in the sternal body and xiphoid fissures also come under this heading [9]. Some surgeons refer to superior cleft sternum as a partial ectopia cordis, but the heart is actually in a normal anatomic position [15–18].

Complete cleft sternum, also called bifid sternum or sternal fissure, is the rarest type, and the sternal bars are completely separate. There may also be diastasis of the rectus muscles. It has been diagnosed by ultrasonography at 21 weeks' gestation [19]. Some authors have reported complete cleft sternum in the literature [19–26].

Inferior cleft sternum is also an incomplete defect: the upper sternum is fused but there is a gap inferiorly. This type may be associated with other abnormalities of midline fusion [15, 16, 18]. In Cantrell's pentalogy, a syndrome commonly described under the classification of abdominal ectopia cordis and characterized by a cleft or absent distal sternum, a crescentic ventral diaphragmmatic defect, a midline ventral abdominal defect with an omphalocele, a defect of the apical pericardium with free communication into the peritoneal cavity, and cardiac malformations, often including either a ventricular septal defect or a ventricular aneurysm [7, 23].

Conclusions

Our case report represented major congenital chest wall deformity, which is a rare disorder in which there is a gap in the midline of the anterior chest wall between the two halves of the sternum, and absent processus xiphoideus in an adult male not associated with cardiac anomalies. In addition to inferior sternal cleft, we noted anatomically important variations in sternum and costae in according to skeletal levels and its costosternal joints. This case is rather unusual as regards the association of an inferior sternal cleft. Especially, thoracic surgeons and radiologists should not have forgotten to encounter.

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