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Case Report

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A RARE CASE REPORT - ISOLATED ASYMPTOMATIC ABSENT MANUBRIUM STERNII IN HEALTHY INFANT

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ABSTRACT

Congenital sternal defects are rare deformities frequently associated with other anomalies of the chest wall and other inner organs. The etiology is not well understood. Congenital abnormalities of the anterior thoracic wall comprise a spectrum of deformities such as thoracic ectopia cordis,cervical ectopia cordis, thoracoabdominal ectopia cordis, pectus excavatum, pectus carinatum, cleft sternum, and short sternum.[1]We report a case of isolated absent manubrium sternii in a 1 month old baby. This combination of clinical features has been described as incomplete presentation of the sternal malformation/vascular dysplasia syndrome. However there is an overlap with features of PHACES syndrome. The combination of clinical features presented in the case has not been mentioned in literature till date.

1. INTRODUCTION

Congenital sternal defects are rare deformities frequently associated with other anomalies of the chest wall and other inner organs like heart. The etiology of these defects is not well understood.[1]

We observed a type of isolated sternal defect to our knowledge not previously reported in literature, the complete absence manubrium sternii were identified, in an asymptomatic infant without inner organs anomalies.

2. CASE PRESENTATION

A 1 month old baby girl was brought to the OPD with history of swelling in the suprasternal region on crying and coughing. She was first order female child born out of non-consanguineous marriage. Mother had a normal antenatal period followed by a normal vaginal delivery at term gestation with a birth weight of 3.1 kg and baby cried immediately after birth and later on normal postnatal period. On examination baby had a depression in the middle of the upper chest which bulged out like a balloon when the baby cried and hyperpigmented dimple. (Figure 1)

Because of suspicion of defect in the sternum investigations were done. Chest X-ray showed a rounded translucency 1.5 cm in size is seen in the midline at cervico-thoracic junction probably represents defect in sternum.

Computer tomography confirmed absence of manubrium sternii.(Figure 2)The remaining skeletal survey was normal. USG shows a homogenously echogenic structure deep to the defect probably represents thymus and intrathoracic and abdominal structures were reported to be normal. Echo-cardiography ruled out valve abnormalities and large vessel defect. The patient was completely asymptomatic and she did not need any medical or surgical treatment.

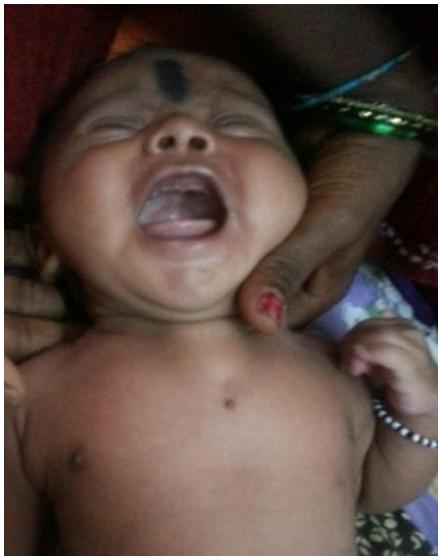

FIGURE 1: Clinical picture of the patient

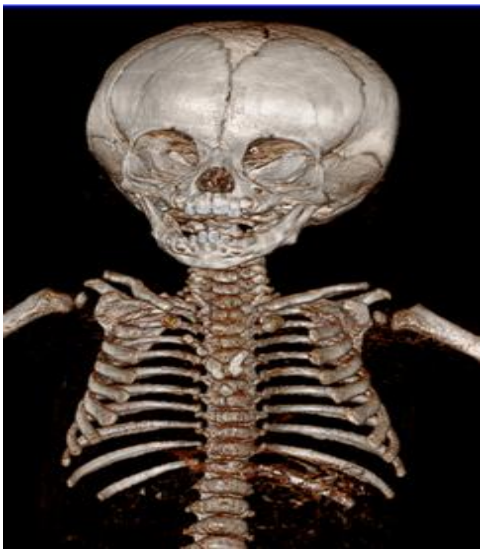

FIGURE 2: The computed tomography scan with three-dimensional reconstruction documents the absent manubrium sternii

3. DISCUSSION

Isolated sternal defect is extremely rare and although exact incidence unknown, it is rarer than Cantrell's pentology whose incidence is 1 in 100,000 live births [(18)] .Although pectus excavatum, pectus carinatum, and cleft sternum can present as isolated deformity and in most cases they are associated with heart and inner organs anomalies and described as symptoms of syndromes like Marfan syndrome, Noonan syndrome, Poland anomaly, and Cantrell pentalogy.[1–7]

In contrast, the etiology of an isolated defect is not well understood. Many factors have been associated with murine models, like alcohol, riboflavin and methylcobalamin deficiency, and HOX4 gene disruption, but no significant associations have been reported in humans.[2–7] The sternum is derived from paired concentrations of thoracic lateral bands that fuse in a cranio-caudal direction by the ninth week of gestation. Disturbances of normal ventral midline thoracic fusion can present as a spectrum of abnormalities, including a prominent suprasternal notch, irregularities in shape of the xiphoid, ectopia cordis, superior sternal cleft or complete sternal cleft.[18,19,20] Antenatal ultrasound in the hands of an experienced sonologist yields the desired information on sternal anatomy and helps to rule out associated developmental anomalies. The 2nd trimester is the best period for sternal study because of absence or small number and size of the ossification centers.Because this is a rare condition, sonographers should keep it in mind as a differential diagnosis in the presence of chest and abdominal wall abnormalities. In addition, 3D sonography is an important complementary tool for diagnosing fetal malformations that are difficult to characterize with 2D sonography and clarifying the quality of subtle fetal movements.

A case that is worth mentioning is that of Santa Rosa da Viterbo, in which a careful study of the well-preserved body showed that she was suffering fromtotal agenesis of the sternum without other apparent malformations.[8, 9] Although there is a large literature about the sternal defects and some reported cases of syndromic short sternum (trisomy 7, trisomy 18, trisomy 9, trisomy 12p, Cantrell pentalogy, Mobius syndrome, and Turner syndrome)[10–17] after a thorough review, we did not find other cases of isolated absent manubrium sternum; therefore, to our knowledge, this is the first case reported in literature.

4. CONCLUSION

It is important to diagnose abnormal sternum prenatally and to rule out other possible associated malformations so that parents are counselled properly and delivery is planned in a tertiary centre with adequate neonatal care.

Early recognition of sternal abnormalities is imperative to avoid life threatening trauma to vital structures. It also markedly influences the choice of repair and prognosis as delayed recognition may result in complex reconstruction with cardiac and vascular compromise

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