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Case Report

Paediatrics Section

Isolated Partial Sternal Agenesis in a Newborn: A Case Report

RS SUSHMA[1], T HARIHARASUDHAN[2], MG RAVANAGOMAGAN[3]

ABSTRACT

Anterior chest wall skeletal anomalies such as sternal cleft or agenesis are rare congenital defects resulting from incomplete fusion of the paired sternal bars during embryogenesis. They may occur as complete or partial defects and can be associated with intracardiac, diaphragmatic, or midline anomalies. The estimated incidence is less than 0.15% of chest wall malformations. Early recognition in the neonatal period is important due to implications for respiratory mechanics, risk of mediastinal injury, and timing of surgical repair. We report a case of isolated partial sternal agenesis in a 35-week preterm male neonate, born small for gestational age (1.9 kg), who developed respiratory distress within the first hour of life. Examination revealed a soft midline depression over the upper chest without pulsation or skin changes. Imaging demonstrated absence of the manubrium and upper sternebrae without mediastinal herniation. Echocardiography, cranial ultrasonography, and chromosomal microarray were normal. The respiratory distress resolved within 24 hours, consistent with transient tachypnoea of the newborn. The infant was managed conservatively with favourable outcome.

Keywords: Chest wall, Congenital abnormalities, Infant, Premature, Sternum abnormalities, Tomography, X-ray computed

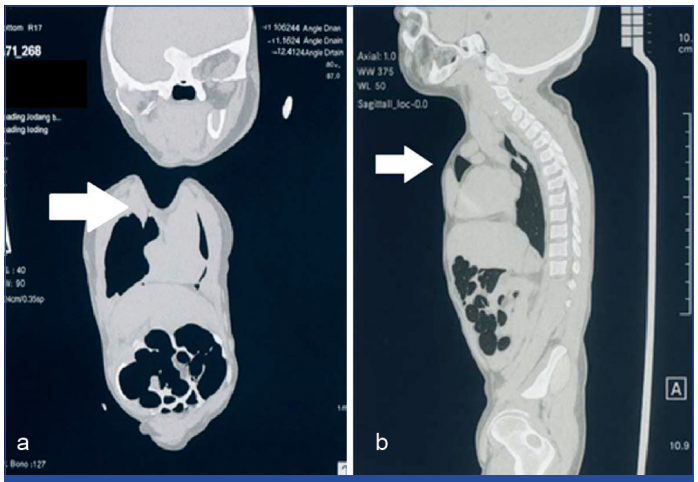
CASE REPORT

A male neonate was delivered at 35 weeks of gestation via spontaneous vaginal delivery to a 28-year-old healthy primiparous non-consanguineous mother. The antenatal course was uneventful, and routine antenatal ultrasounds showed no anomalies. At birth, the baby weighed 1.9 kg (below the 10th centile for 35 weeks), with length and head circumference at the lower percentile. APGAR scores were eight and nine at one and five minutes, respectively. Within the first hour of life, the baby developed respiratory distress characterised by tachypnoea with a respiratory rate of around 70 per minute, moderate intercostal retractions, nasal flaring, and mild desaturations requiring supplemental oxygen via hood. On examination, a soft midline depression was noted over the upper chest in the manubrial region [Table/Fig-1] without visible pulsation, overlying skin changes, herniation of mediastinal structures, or paradoxical motion. Heart sounds were normal, peripheral pulses were symmetrical, and there were no abdominal wall, scalp, or limb anomalies. The rest of the systemic examination was unremarkable. The infant was admitted to the neonatal intensive care unit for respiratory support and received supplemental oxygen, thermal care, and routine newborn care. Over the next 24 hours, respiratory distress resolved, oxygen was discontinued, and a diagnosis of transient tachypnoea of the newborn was made. However, due to the chest wall abnormality, skeletal imaging was taken. Chest radiography revealed absence of the manubrium and upper sternum with normally aerated lung fields and no mediastinal herniation [Table/Fig-2]. Computed tomography of the thorax confirmed isolated partial sternal agenesis involving the manubrium and first two sternebrae [Table/Fig-3a,b,4] with intact lower sternum, ribs, and clavicles, and no paradoxical motion or mediastinal protrusion. Echocardiography [Table/Fig-5] demonstrated a structurally normal heart without intracardiac or great vessel anomalies. Cranial ultrasonography [Table/Fig-6] was normal, excluding associated central nervous system defects.

Routine haematological and biochemical parameters were within normal limits, and chromosomal microarray analysis revealed no pathogenic variants. In a neonate presenting with a midline anterior chest wall defect, differentials include partial or complete sternal cleft or agenesis, Cantrell's pentalogy, PHACE syndrome, scalp or aplasia cutis of the scalp with bone defect, anterior chest wall trauma, or isolated sternal hypoplasia or bifid sternum. In this case, the absence of cardiac, diaphragmatic, or pericardial anomalies and the imaging findings supported isolated partial sternal agenesis. As the respiratory distress resolved and was attributed to

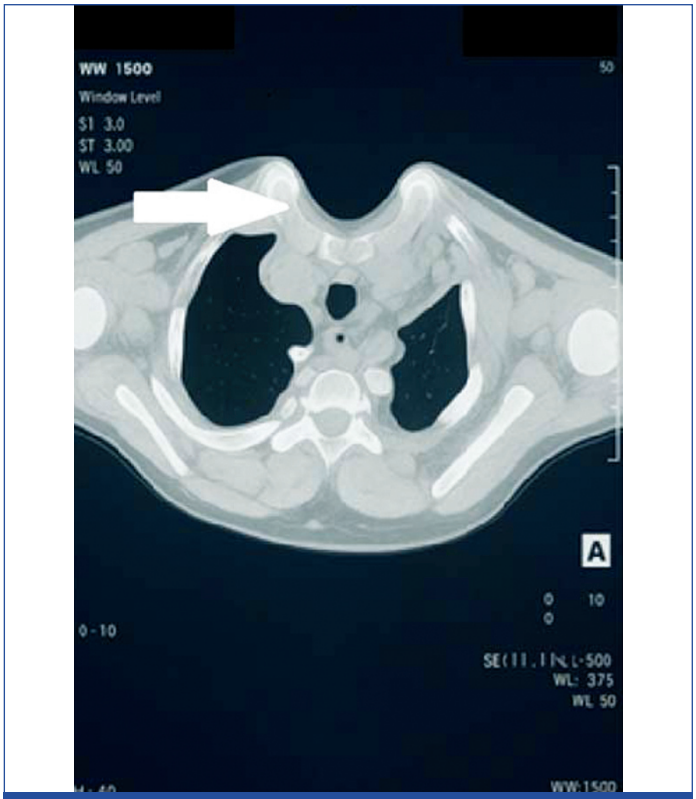
[Table/Fig-1]: External appearance of the neonate showing a soft midline chest wall depression.

[Table/Fig-2]: Chest radiograph showing absence of manubrium and upper sternum.

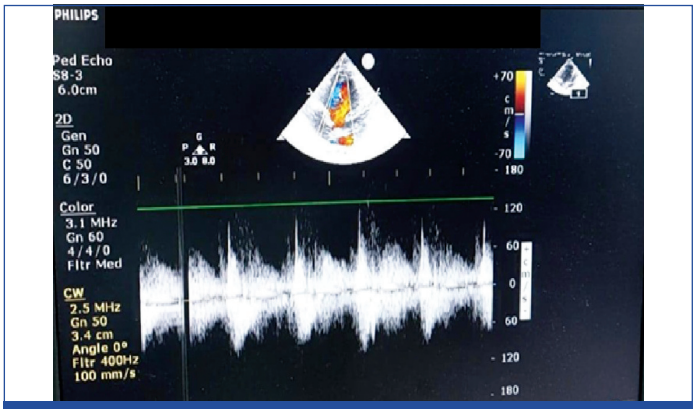


[Table/Fig-3]: a) Coronal CT image showing absence of the manubrium and upper sternum without mediastinal herniation; b) Sagittal CT image confirming upper sternal defect without mediastinal protrusion.

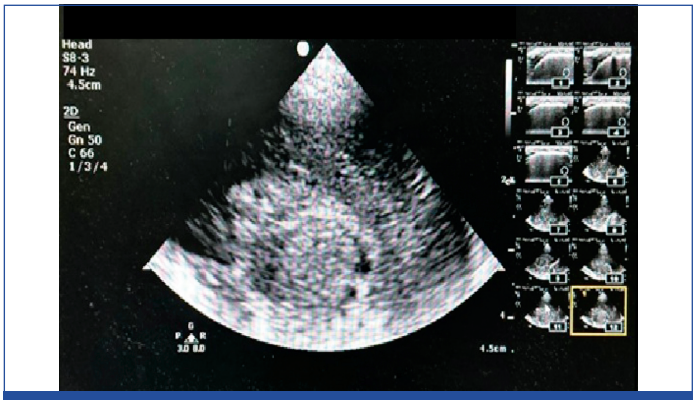
transient tachypnoea of the newborn, no surgical intervention was indicated during the neonatal period. The neonate was managed conservatively with supportive care, monitoring of respiratory status, thermoregulation, and feeding support. Given the chest wall defect, periodic follow-up was advised with paediatric and thoracic surgery teams for evaluation of chest wall dynamics, respiratory function, and growth. The parents were counselled about the anomaly, possible future need for surgical repair if required, and the importance of protecting the chest from trauma. The neonate remained stable, was weaned off oxygen within 24 hours, tolerated full feeds, and was discharged home on day five of life in good condition. At follow-up at two months of age, the infant was thriving with a weight of approximately 3.2 kg, no respiratory symptoms, and an unchanged chest wall without paradoxical movement. Further follow-up is planned to monitor development, chest wall growth, and any evolving clinical sequelae.



[Table/Fig-4]: Axial computed tomography image at the level of the thoracic inlet (approximately T2-T3 vertebral level) showing absence of the manubrium with intact clavicles and no evidence of mediastinal herniation.



[Table/Fig-5]: Echocardiography showing a structurally normal heart with no evidence of intracardiac or great vessel anomalies.



[Table/Fig-6]: Cranial ultrasonography demonstrating normal brain structures with no evidence of associated central nervous system defects.

DISCUSSION

Partial sternal agenesis is an exceedingly rare congenital anterior chest wall anomaly resulting from failure of midline fusion of the paired sternal bars during the 7th-10th weeks of gestation [1,2]. Earlier surgical experience demonstrated that neonatal chest wall compliance allows primary approximation of sternal bars with favourable outcomes when repaired early in life [1]. Subsequent

descriptions of midline developmental anomalies further highlighted the embryological basis and potential syndromic associations of sternal defects [2].

Larger reviews have detailed the phenotypic spectrum ranging from small V-shaped clefts to complete absence of the sternum and emphasised the importance of careful evaluation and planning [3]. Complete congenital sternal clefts with mediastinal exposure requiring early repair have also been reported [4].

Primary closure in the neonatal period is generally preferred because increased chest wall elasticity permits tension-free approximation and reduces the risk of mediastinal compression [5]. Isolated congenital absence of the sternum without associated anomalies remains distinctly uncommon [6], although neonatal cases with visible cardiac pulsation or respiratory compromise have been described [7]. Surgical repair in early infancy has shown favourable outcomes in selected cases [8], while recent reports of isolated V-shaped defects support individualised management depending on symptom severity [9].

Importantly, sternal defects may occur as part of broader syndromic associations. They are classically described in Cantrell's pentalogy, which comprises a lower sternal defect, anterior diaphragmatic defect, pericardial defect, midline supraumbilical abdominal wall defect, and intracardiac anomalies. Sternal clefts have also been reported in association with PHACE syndrome, reflecting shared disturbances in midline embryological development [2,10]. These associations necessitate thorough cardiac and systemic evaluation in all affected neonates. In our patient, echocardiography and imaging excluded such anomalies, which likely contributed to the favourable course. Recent surgical techniques have further demonstrated successful outcomes even in complex cases of congenital sternal absence [11]. The successful deployment of modern alternative/advanced techniques (such as prosthetic meshes, specialized autologous grafts, or customized biological flaps) specially to resolve complex or late - presentation variations. Additionally, follow-up data from recent case reports indicate good intermediate-term outcomes after repair, supporting early intervention strategies [12].

In the present case, the antenatal course was uneventful and routine obstetric ultrasonography did not detect the defect. Prenatal detection of isolated sternal anomalies can be challenging because standard anomaly scans focus primarily on intracardiac structures and major organ systems; small anterior chest wall defects may be overlooked due to fetal position, acoustic shadowing from ossification centres, limited resolution, and absence of mediastinal protrusion. Clinically, the defect presented as a soft midline depression without paradoxical chest wall motion or visible pulsation. The transient respiratory distress resolved within 24 hours and was consistent with transient tachypnoea of the newborn rather than persistent respiratory failure, suggesting that the defect did not significantly impair respiratory mechanics. The absence of mediastinal herniation or intracardiac anomaly likely contributed to the favourable clinical course.

Prognosis in isolated partial defects without associated anomalies is generally excellent; however, long-term surveillance is advisable to monitor chest wall growth and the potential need for delayed intervention.

Several paediatric case reports have described isolated sternal defects with variable presentation and management.

Basraoui D et al., reported a 20-day-old newborn with an isolated sternal cleft confirmed on CT who underwent early surgical repair with favourable postoperative recovery [13]. Powar RS et al., described an eight-year-old child with an isolated cleft sternum who underwent primary surgical closure with good cosmetic and functional outcome [14].

This case highlights that isolated partial sternal agenesis without cardiopulmonary compromise can be safely managed conservatively with careful follow-up.

CONCLUSION(S)

This case highlights that isolated partial sternal agenesis in a preterm neonate may present as a subtle chest wall defect without significant impact on respiratory mechanics and can coexist with transient tachypnoea of the newborn. In the absence of mediastinal herniation or associated anomalies, conservative management with careful follow-up is a safe and effective approach.

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PARTICULARS OF CONTRIBUTORS:
1. Junior Resident, Department of Paediatrics, Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India.
2. Assistant Professor, Department of Paediatrics, Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India.
3. Associate Professor, Department of Paediatrics, Sree Balaji Medical College and Hospital, Chennai, Tamil Nadu, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
MG Ravanagomagan,
G-4, R Block, SBMCH, No. 7, CLC Works Road, Chrompet,
Chennai-600044, Tamil Nadu, India.
E-mail: drmgr04@gmail.com

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