A Case of Mesenchymal Hamartoma of the Chest Wall in a 4-Month-Old Infant

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Patient: Male, 4-months
Final Diagnosis: Mesenchymal hamartoma
Symptoms: Asymptomatic chest wall mass
Medication: —
Clinical Procedure: —
Specialty: Radiology

Objective: Rare disease

Background: Mesenchymal hamartoma of the chest wall is a rare benign lesion that typically presents in early infancy. However, the clinical presentation can be atypical, with imaging features that mimic infection or malignancy. Imaging combined with histopathology is essential in the diagnosis. A case of mesenchymal hamartoma of the chest wall in a 4-month-old infant is presented.

Case Report: A 4-month-old infant had an incidental finding of a large right-sided chest wall mass. Initial imaging included thoracoabdominal ultrasound (US), computed tomography (CT), and magnetic resonance imaging (MRI). Histology of an initial open biopsy was inconclusive. The diagnosis of mesenchymal hamartoma was confirmed by histology of the resection specimen, which showed a benign, mixed, chondroid, mesenchymal, and cystic hamartoma with areas of calcification and ossification.

Conclusions: This case showed that the diagnosis of mesenchymal hamartoma of the chest wall, which is characterized by heterogeneous components, may require a combined approach for the diagnosis that includes imaging and histology. Increased clinical awareness of mesenchymal hamartoma in infants may help to guide the approach to the correct diagnosis and prevent unnecessarily radical treatment for this benign condition.

MeSH Keywords: Diagnostic Imaging • Hamartoma • Mesenchymal Stromal Cells • Pediatrics • Thoracic Wall

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/913228
Background

Mesenchymal hamartoma of the chest wall, also known as chondromesenchymal hamartoma, is a rare benign lesion of early infancy that arises from one or more ribs [1,2]. Mesenchymal hamartoma is heterogeneous and consists of cartilage and mesenchymal cells and can show cystic change, calcification, and ossification, and grows by expansion [3,4]. Combined imaging and histology may be required to confirm the diagnosis. This report describes the clinical and radiological features and the approach to the treatment of a 4-month old with an incidental finding of a mesenchymal hamartoma of the chest wall.

Case Report

A 4-month-old previously well infant presented with a history of several days of poor feeding, with no fever. Physical examination was unremarkable and did not identify any palpable masses. Lung air entry was symmetrical. Initial laboratory investigations including urinalysis suggested a diagnosis of cystitis and the patient commenced empirical treatment with antibiotics. He was evaluated with ultrasound (US) imaging of the kidneys and urinary bladder that showed a large, heterogeneous, vascular, and predominantly solid mass located at the right lateral thoracoabdominal junction adjacent to the liver (Figure 1A). The mass was surrounded by hypoechoic pleural fluid and impinged upon the superior aspect of the right hemidiaphragm, which indicated an intrathoracic origin (Figure 1B). There was inferior displacement of the right lobe of the liver and right kidney. A few echogenic calcific foci and hypoechoic cystic spaces were noted within the mass (Figure 1C). The kidneys and urinary bladder were normal.

Magnetic resonance imaging (MRI) was then performed. Coronal T2-weighted MRI supported the US findings of a large well-defined lobulated right intra-thoracic mass (Figure 2A). MRI showed that the mass appeared to arise from the posterolateral aspect of the expanded right 8th rib. The origin of the mass was identified more clearly in the sagittal plane as the mass was surrounded by curvilinear thin T2-weighted hypointensity that represented the cortical margins of the rib, which were not eroded (Figure 2B). There was bone remodeling of the adjacent 7th and 9th ribs.
Figure 2. Coronal, sagittal, and axial T2-weighted magnetic resonance imaging (MRI) of the thoracoabdominal wall in a 4-month-old infant with a right-sided mesenchymal hamartoma of the chest wall. (A) Coronal T2-weighted magnetic resonance imaging (MRI) supports the ultrasound (US) findings of a large, well-defined, lobulated, right intrathoracic mass that displaces the right kidney (RK) inferiorly. (B) Sagittal T2-weighted MRI shows thin curvilinear T2-weighted hypointensity surrounding the mass (arrows), which represent the preserved cortical margins of an expanded rib, indicating that the mass arises from the rib. (C) Axial T2-weighted fat-saturated MRI shows a lobulated endophytic component of the mass with fluid-fluid levels in keeping with an aneurysmal bone cyst component.

On T1-weighted and T2-weighted MRI, hyperintense components were present that were interpreted as blood-filled cysts. The mass had a large endophytic component and was multilobulated with multiple fluid-fluid levels in keeping with an aneurysmal bone cyst (ABC) component (Figure 2C). There was significant mass effect with partial atelectasis of the middle and lower lobes of the right lung and leftward shift of the heart. The right hemidiaphragm, the right lobe of the liver, and the right kidney were displaced inferiorly. There was no intraperitoneal extension of the mass. Limited non-enhanced computed tomography (CT) images of the thorax were acquired, which confirmed the origin of the mass from the rib. The mass also showed internal areas of ossification (figure 3).

The patient then underwent an open biopsy of the rib mass which showed features suggestive of an aneurysmal bone cyst (Figure 4A). As the open biopsy was considered to be only partially representative of the imaging findings, the patient subsequently underwent surgical resection of right chest wall mass, including the right 7th and 8th ribs. Histopathology of the resection specimen confirmed the diagnosis of a mesenchymal hamartoma and showed the typical features of chondroid,
mesenchymal, and vascular elements, as well as calcification and ossification (Figure 4B).

Post-operative follow-up at one month and three months showed the patient to be clinically and developmentally well with no evidence of recurrence or scoliosis.

Discussion

In 1979, Mcleod and Dahlin proposed the term mesenchymal hamartoma [1]. Mesenchymal hamartoma is an extremely rare condition with an estimated incidence of less than one per million population [2]. Mesenchymal hamartoma of the chest wall presents in neonates and infants and can be associated with a chest wall deformity or mass and may present with symptoms of respiratory distress [3,4]. A subclinical presentation of mesenchymal hamartoma of the chest wall may also occur, as in the present case, in which the finding was incidental. Due to the non-specific clinical presentation, the radiological appearance and the histological features seen on small needle biopsies may be suspicious for infection or malignancy [5–7].

The characteristic features of mesenchymal hamartoma on chest radiographs are those of an ossified intrathoracic soft tissue mass with cortical thinning that may deform the affected rib [3]. On ultrasound (US), a heterogeneous solid and cystic mass containing echogenic foci with ossification may be found. On computed tomography (CT) imaging, an ossified solid and cystic mass with single or multiple rib involvement may be shown [3,8]. Solid and cystic masses with fluid levels have been described on magnetic resonance imaging (MRI) of mesenchymal hamartoma of the chest wall [3]. MRI may also show areas of T1-weighted hypointensity indicating the presence of chondroid elements, allowing the differentiation of the mass from an aneurysmal bone cyst [9].

On US imaging alone, it can be challenging to evaluate a large mass in a young infant. When a large mass is seen on US, it is equally important to visualize the edge of the mass and its...
relations with surrounding structures, notably the diaphragm, which may enable the radiologist to determine whether the mass is intrathoracic or intraperitoneal in location. In the patient in this report, the mass was surrounded by hypoechoic pleural fluid, which established an intrathoracic (supradiaphragmatic) location rather than an intraperitoneal location. Imaging was important to exclude a possible adrenal mass, for example, due to neuroblastoma.

This case demonstrated the possible challenges in making the diagnosis of mesenchymal hamartoma of the chest wall, and this case was unusual in that a chest radiograph was not performed prior to US or MRI. The origin of the lesion was identified using the sagittal plane on MRI by carefully examining the T2-weighted hypointense bone cortex of the rib. This approach helped the radiologist to determine that the lesion originated from the rib rather than the pleura.

Therefore, as this case has shown, the diagnosis of mesenchymal hamartoma of the chest wall can be made in a neonate or young infant when there is a mass arising from the ribs that contains cystic spaces and internal ossification on cross-sectional imaging [4]. Without imaging and diagnostic histopathology, the differential diagnosis of mesenchymal hamartoma of the chest wall can include a primary Ewing sarcoma or chest wall metastases from neuroblastoma, leukemia, and lymphoma. However, these malignant tumors do not show cystic components and metastatic tumors in infancy are less likely to show extra-thoracic involvement [4]. The common benign rib lesions that may be included in the differential diagnosis are fibrous dysplasia, hemangioma, and Langerhans cell histiocytosis (histiocytosis-X), which do not show internal ossification [4].

Combined positron emission tomography (PET) with CT imaging have a limited diagnostic role in mesenchymal hamartoma, which shows low hypermetabolic fluorodeoxyglucose (FDG) uptake that can mimic malignancy [10]. Where clinical or imaging features are atypical, histopathology is required for definitive diagnosis. In this case, a biopsy of the rib mass showed features more in keeping with an aneurysmal bone cyst, which was interpreted to be the result of sampling error. Histopathology of the completely excised mass confirmed the diagnosis of benign mesenchymal hamartoma. This case has shown that when a definitive histopathological diagnosis is required without complete excision, a percutaneous biopsy under US or CT guidance should aim to target the more solid or ossified components, rather than the cystic areas, to improve diagnostic accuracy.

The accuracy of early diagnosis of benign mesenchymal hamartoma of the chest wall is important, as in cases where chemotherapy has been initiated for the treatment of a presumed diagnosis of embryonal sarcoma, chemotherapy has been reported to result in patient mortality [11,12]. The treatment of mesenchymal hamartoma in symptomatic patients is surgical resection, which results in favorable prognosis [5]. Previous cases reports of spontaneous regression raise the possibility of a conservative treatment approach especially for smaller asymptomatic cases of mesenchymal hamartoma of the chest wall [13,14].

Conclusions

Mesenchymal hamartoma of the chest wall is a rare benign lesion that can arise from the rib and is more common in infants. The clinical presentation and imaging findings may initially raise concern for malignancy, as the lesion may impinge on neighboring structures. As this case has shown, histopathology can confirm the classical features of benign mesenchymal hamartoma and may be required to exclude the diagnosis of aneurysmal bone cyst or malignancy. Also, this case has shown that percutaneous biopsy without imaging guidance may be an initial approach to diagnosis, but due to the heterogeneous nature of mesenchymal hamartoma, may not provide a definitive diagnosis. Clinical awareness of mesenchymal hamartoma of the chest wall may guide the approach to confirmatory diagnosis and the most appropriate treatment.

Statement

Ethics approval for this case report was waived by the Singhealth Centralized Institutional Review Board (CIRB).

Conflict of interest

None.

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