Letter to the Editor

Pioneering prenatal ultrasonic diagnosis of fetal mediastinal teratoma: a comprehensive case description unveiling diagnostic nuances

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Introduction

Mediastinal tumors exhibit specific locations, including superior, anterior, middle, and posterior regions, based on their histological characteristic (1). Teratomas, classified as germ cell tumors, commonly originate in the anterior mediastinum. Notably, early detection of mediastinal tumors through prenatal fetal ultrasound is achievable (2). In this context, we present a case of a fetal mature cystic mediastinal teratoma, confirmed through ultrasonography and histopathology. This case underscores the notion that early diagnosis of fetal mediastinal teratomas can often be accomplished through prenatal echocardiography.

Case presentation

A 25-year-old woman was referred at 35 weeks of gestation due to the discovery of a cystic mass in the right anterior mediastinum of the fetus on an obstetric ultrasound scan. The patient had no history of medical, surgical, or familial diseases. She was admitted to our hospital following a 1-week history of elevated blood pressure without an obvious cause. On the same day, a detailed ultrasonographic (US) examination, utilizing a GE Voluson E10 scanner (GE Healthcare, Chicago, IL, USA), was conducted. A routine prenatal ultrasound revealed a heterogeneous, lobulated mass in the fetal chest above the heart, measuring 37×36×40 mm (*Figure 1A,1B*). The cyst displaced the mediastinum and heart to the left. Color Doppler sonography indicated no blood flow in the cyst (*Figure 1C*). Fetal biometric measurements were appropriate for gestational age, and no other chest or cardiovascular abnormalities were identified. There was no large blood vessel compression, no lung masses or discrete connection of the mass to the heart. The remaining fetal anatomical tests, including spinal column (hemivertebra absence), amniotic fluid volume, fetal stomach size, biophysical characteristics, fetal umbilical cord, and middle cerebral artery Doppler were normal. No fetal hydrops were present, although the thymus was not clearly assessed. Throughout the third trimester, fetal growth rates and biophysical characteristics remained normal.

The patient underwent spontaneous delivery of a neonate at 39 weeks of gestation, with Apgar scores rated 9 and 9 at 1 and 5 minutes, respectively, and a birth weight of 2,145 grams. No neonatal complications were observed.

Post-birth, a chest computed tomography (CT) scan revealed a large, low-density lesion in the anterior mediastinum, a fluid-filled, well-demarcated cyst with a 4 cm anterior-posterior component (*Figure 2A*), and contrast-enhanced CT (*Figure 2B*) showed that the mass had no enhancement. Magnetic resonance imaging (MRI)

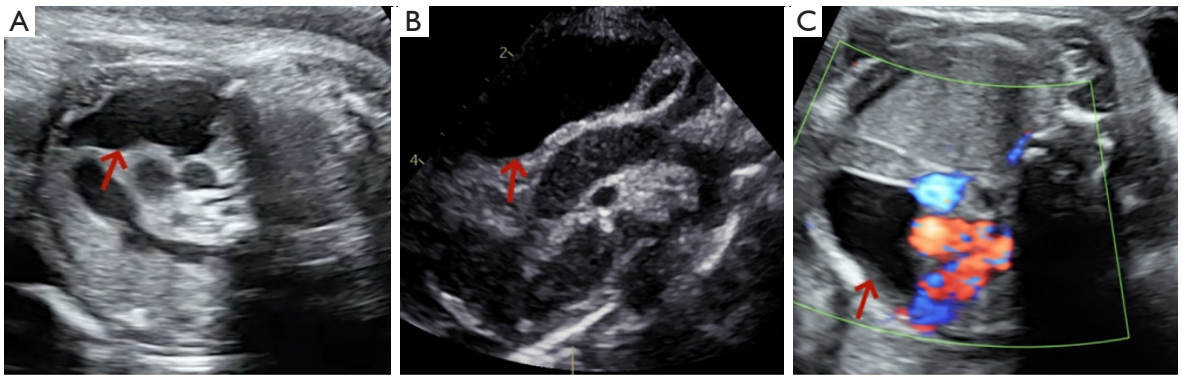

Figure 1 Ultrasound examination. Ultrasound examination (A,B) revealed a hypoechoic, avascular, single-compartment cystic mass (red arrows) in the right posterior chest of the fetus. Color Doppler ultrasound (C) showed no blood flow signal. The cystic mass is showed by red arrow.

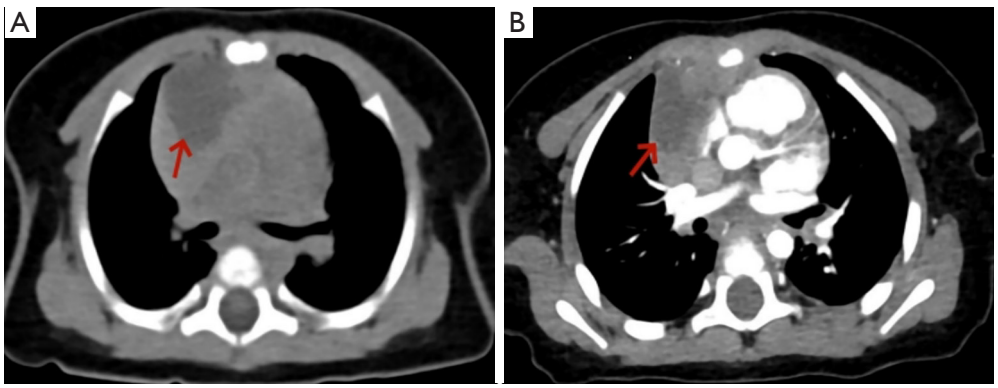

Figure 2 Mediastinal window on axial chest CT. Axial chest CT (A) demonstrated the presence of a low-density anterior mediastinal mass (red arrow). Contrast-enhanced CT (B) showed the mass (red arrow) has no enhancement. CT, computed tomography.

showed an encapsulated mass with high signal intensity on T2-weighted imaging, which contained fluid components (*Figure 3A*,*3B*). Follow-up echocardiograms at 1, 6, and 12 months after birth showed no noticeable changes in measurement, exterior condition, or impact of the cyst on neighboring structures.

Surgical intervention was performed on the child at 12 months of age. The cystic was mass removed intact; it was a solid grayish white and yellow color with no immature or malignant components. Tissue analysis confirmed that it was a mature teratoma with a cystic cavity containing yellow fluid and hair. Fine adhesions to the pericardium and thymus were easily dissected, leaving these structures intact. The encapsulated lobulated mass measured 9×5×2 cm, displacing and compressing the heart but not directly involving the thymus or invading lungs or blood vessels (*Figure 4*).

Under hematoxylin-eosin staining, the solid component of the mass showed features indicating a high likelihood of a mature cystic teratoma located in the anterior mediastinum, indicating that all complex elements were mature (*Figure 5*).

The patient experienced no major complications and was discharged 1-week post-operation. Subsequent follow-up revealed no signs of local recurrence.

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this article and accompanying images. A copy of the written consent is

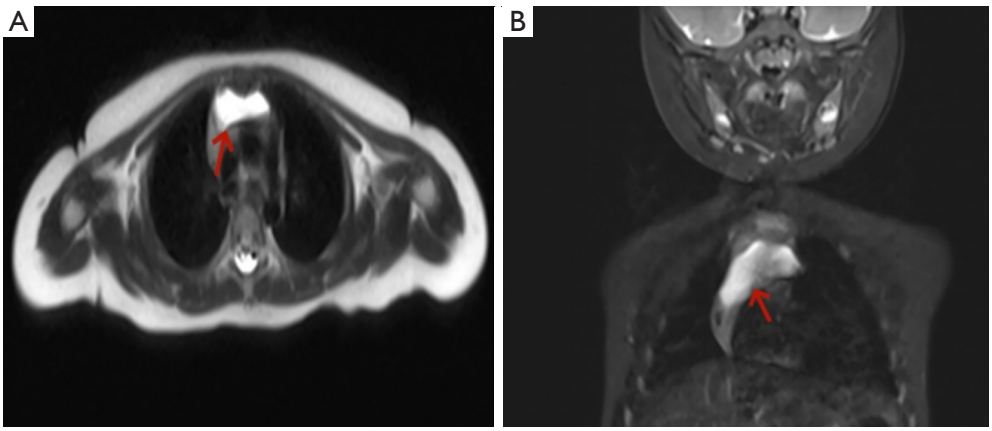

Figure 3 Sectional and coronal T2-weighted MR images. The cyst (red arrows) produced a homogeneous, hyperintense signal (A,B), clearly localized within the right anterior mediastinum. The MR findings are consistent with a cystic mass (red arrows). MR, magnetic resonance.

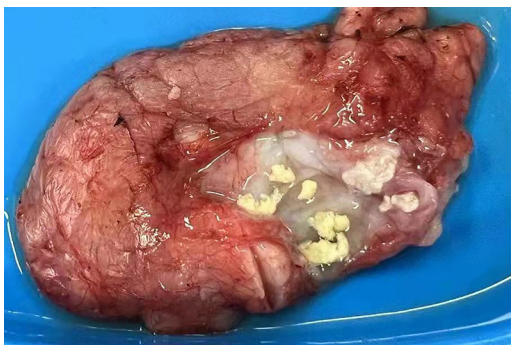

Figure 4 Gross examination of the specimen. Upon gross examination of the specimen, the tumor was identified as cystic.

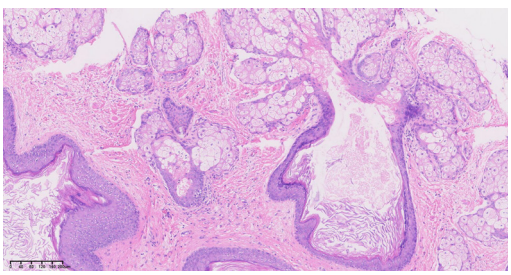

Figure 5 Histological examination. Microscopic appearance of the mass under hematoxylin-eosin staining revealed the presence of collagen fiber, pilosebaceous units, stratified squamous epithelium, and fatty tissue. This tumor was diagnosed as a mature cystic teratoma without malignancy, with no immature elements observed.

available for review by the editorial office of this journal.

Discussion

Fetal teratomas in the field of fetal medicine are infrequent occurrences, typically manifesting in the anterior mediastinum as either a median or paramedian mass (3). These teratomas originate from totipotent cells during embryonic development, and their pathogenesis in extragonadal sites is associated with the displacement of primitive germ cells in the course of embryogenesis. Among congenital tumors, teratomas rank as some of the most common, accounting for 16.6% of all fetal tumors, with an incidence of 1 in 20,000–40,000 live births (4,5). These tumors are categorized into mature teratomas, which contain differentiated tissue and immature teratomas from all 3 germ layers, in addition to neural ectodermal components, ventricular rosettes, and immature mesenchyme. Immature teratomas account for only 1% of cases, and surviving cases in the perinatal period without postoperative complications are sparse (6). The case presented here involves a large cystic mature mediastinal teratoma that was prenatally identified at 35 weeks of gestation.

Immature teratomas commonly exhibit associations with hydrops placentae, polyhydramnios, and severe hydrops fetalis characterized by generalized skin edema, ascites, and pleural effusions. In contrast, mature teratomas constitute the most prevalent type of mediastinal germ cell

tumor (7). These tumors are typically cystic and often remain asymptomatic unless they attain a considerable size, leading to the compression of vital organs (8). On anatomical specimens, mature teratomas appear as ball-shaped, lobed, encapsulated, and multicystic tumors with a composition ranging from lipid-rich fluids and cheese-like substances to well-structured tissues such as teeth and hair (9-11).

Mediastinal mature teratoma is a rare, benign, slow-growing tumor that occurs primarily in or near the thymus and accounts for 75% of primary germ cell tumors of the mediastinum (12). These lesions, identified as large and benign, consist of 1 or more embryonic germ layers (ectoderm, mesoderm, and endoderm) and mirror their gonadal counterparts in terms of histologic features. The embryonic period's first 3 months emerge as a critical stage of development, during which embryos exhibit heightened sensitivity to teratogenic factors. Prenatal diagnosis of mediastinal teratomas can be readily achieved using 2-dimensional ultrasonography, as the mass typically manifests both cystic and solid components, along with calcification spots and acoustic shadows.

When teratomas manifest in the fetal mediastinum, the substantial tumor mass can exert compression on surrounding tissues, potentially impacting fetal lung development. However, in the case presented herein, the fetal lungs exhibited no signs of compression.

Ultrasonography plays a pivotal role in accurate prenatal diagnosis. A meticulous Doppler ultrasound assessment not only aids in the diagnosis of mediastinal teratomas but also facilitates the evaluation of fetal outcomes (13). The 3 major US patterns associated with mediastinal teratomas include mixed masses with heterogeneous echo, homogeneous, and high flowability within solid masses. A complex mass with heterogeneous echogenicity is the most common feature followed by a homogeneous echogenic mass and floating spherules within a cystic mass. In our patient, ultrasonography revealed a homogeneous echogenic cystic lesion with heterogeneous hyperechoic intracellularity located in the anterior mediastinum. Srisupundit's (14) study highlights complex heterogeneous echogenicity as a major sonographic feature of mediastinal teratomas. The presence of diffuse hyperechoic lines and dots in a teratoma, indicative of hair and watery or sebaceous fluid, aligns with the embryologic genesis of mature cystic teratomas.

MRI proved valuable in assessing the tumor's relationship with great vessels and vital structures. In our case, an accurate post-operative diagnosis was primarily obtained through CT and MRI.

Accurate prenatal ultrasound diagnosis and close monitoring of the fetus are important for the treatment modalities utilized at delivery. The treatment plan may include the following 3 modalities: surgery within the uterus, an exutero intrapartum therapy (EXIT) procedure, or surgical resection after birth. Generally, surgical removal results in a favorable prognosis for both mature and immature mediastinal teratomas; in this case, mortality and health status were observed to be good during the different follow-up periods. Although immature teratomas are rare, they can recur, making it necessary to follow up with tumor markers and the like.

The EXIT-to-resection procedure emerges as the optimal approach, allowing immediate fetal thoracotomy and surgical mass removal to prevent acute respiratory distress. The EXIT procedure has been well-described in the past in the management of large intrathoracic and neck masses which compromise airway by compression (13). For cystic lesions in the anterior mediastinum, complete surgical resection, if feasible, remains the best treatment option, offering an excellent prognosis with long-term survival.

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Footnote

Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://qims.amegroups.com/article/view/10.21037/qims-24-492/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this article and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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