

Progressive abdominal distension as an unusual presentation of mediastinal teratoma in an infant: A case report and literature review

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Abstract

Mediastinal masses in pediatric patients often pose a significant diagnostic challenge for both clinicians and radiologists due to their broad spectrum of etiologies, ranging from benign lesions to malignant conditions.

Mediastinal teratomas are germ cell tumors that typically arise in the anterior mediastinum and, less commonly, in the posterior mediastinum. They represent the most frequent form of extragonadal germ cell tumors [1].

We report the case of a 02-year-old female infant with no significant past medical history, who presented with progressive, asymmetric abdominal distension predominantly on the left side, noted over one year by her mother, with preserved general condition.

On physical examination, the patient was in good general condition and afebrile. Abdominal examination revealed a firm left subcostal mass fixed to deep planes, associated with clinical signs suggestive of a left basal pleural effusion.

Chest radiography followed by thoraco-abdominal CT scan revealed a large posterior mediastinal mass occupying nearly the entire left hemithorax. The lesion was mixed solid-cystic, containing fat and scattered calcifications, and caused a significant mass effect with contralateral mediastinal shift and diaphragmatic depression.

Biological investigations showed elevated alpha-fetoprotein (AFP) and negative β -hCG. Initial biopsy suggested a mature teratoma without immature components in the sampled tissue. The patient underwent neoadjuvant chemotherapy followed by complete surgical excision of the mass.

Histopathological examination of the surgical specimen confirmed an immature pluritissular teratoma, grade 1.

The postoperative course was uneventful, with favorable clinico-radiological evolution. The patient is currently under regular follow-up.

Keywords: Mediastinal teratoma; Immature; Mature; Tumor markers

1. Introduction

Teratomas are germ cell tumors derived from the three primitive embryonic germ layers (endoderm, mesoderm, and ectoderm) [2].

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Primordial germ cells, when arrested or abnormally migrating, may proliferate and give rise to a spectrum of lesions ranging from benign forms (mature teratoma and grade 1 immature teratoma) to malignant lesions (high-grade immature teratomas and other malignant germ cell tumors), and more rarely to secondary malignant transformation [3].

They are most often of gonadal origin but may rarely arise outside the gonads, in which case they are referred to as extragonadal teratomas [2].

The mediastinum is the most common extragonadal site, and the majority of teratomas occurring in this location are benign [2].

In infants under one year of age, immature teratomas are relatively common (approximately 40%) and may be detected prenatally [1].

Imaging plays a crucial role in the diagnostic orientation of teratomas. However, the definitive diagnosis relies on histopathological examination obtained through biopsy or surgical resection.

It is therefore essential to distinguish a pure teratoma from lesions containing an additional malignant germ cell tumor component, most commonly a yolk sac tumor. In such cases, the lesion is considered and managed as a mixed malignant germ cell tumor [3].

For this reason, serum tumor markers of secreting malignant germ cell tumors should be systematically assessed prior to any surgical management of a teratoma, particularly alpha-fetoprotein (AFP) for yolk sac tumors and beta-human chorionic gonadotropin (β -hCG) for choriocarcinoma [3].

Therapeutic management differs accordingly: unlike malignant germ cell tumors, pure teratomas are not chemosensitive, and treatment is primarily based on complete surgical excision [3].

2. Case presentation

We report the case of a 2-year-old female infant with no significant past medical history, who presented with progressive and asymmetric abdominal distension, predominantly on the left side, evolving over one year and noted by her mother, with preserved general condition.

On clinical examination, the patient was in good general condition and afebrile. Abdominal examination revealed a firm left subcostal mass fixed to the deep planes, associated with clinical signs suggestive of a left basal pleural effusion.

Chest radiography demonstrated a heterogeneous opacity of fluid density involving the left hemithorax, without obscuration of the cardiac border and without air bronchogram (Figure 1).

Thoraco-abdominal computed tomography (CT) revealed a large, poorly defined left posterior mediastinal mass occupying nearly the entire left hemithorax. The lesion was mixed solid and cystic, containing scattered calcifications and fatty components, measuring approximately $9.5 \times 10 \times 18$ cm (anteroposterior \times transverse \times craniocaudal). It exerted a significant mass effect, displacing the descending aorta, inferior vena cava, azygos vein, and tracheobronchial tree to the right, as well as the diaphragm, left kidney, spleen, and transverse colon inferiorly (Figure 2).

Initially, a diagnosis of neuroblastoma was suspected, and emergency chemotherapy was initiated according to the SMOP-NBL 1 protocol. Meanwhile, a CT-guided biopsy was performed, suggesting a mature teratoma without immature components in the sampled tissue. Tumor marker analysis showed a moderately elevated alpha-fetoprotein (AFP) level (22 ng/mL) and negative β -hCG.

In the absence of baseline AFP measurement prior to chemotherapy initiation, the mass was considered a low-risk malignant germ cell tumor, and neoadjuvant chemotherapy was continued. However, follow-up CT evaluation at the end of treatment showed no significant response, with stable imaging findings.

After multidisciplinary team discussion, surgical management was decided despite the large size and potential morbidity of the tumor, with informed consent obtained from the family.

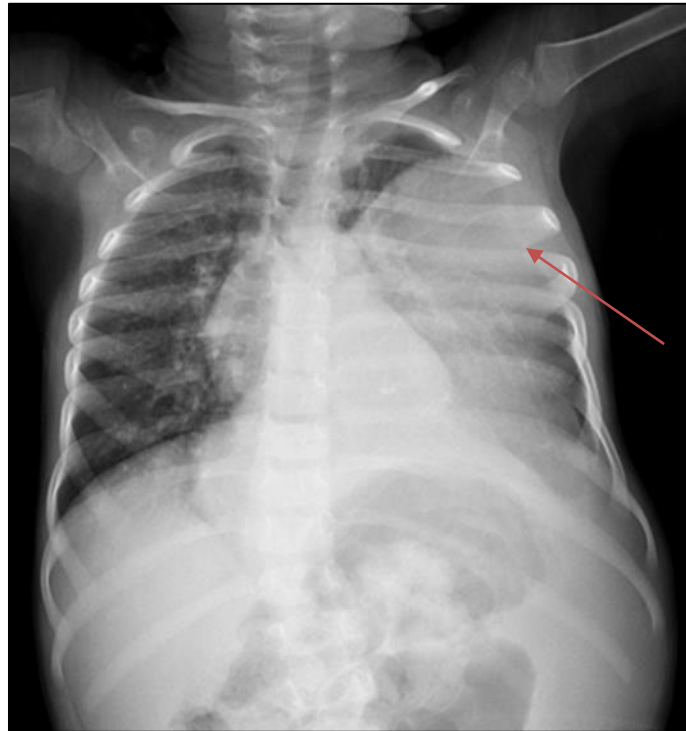
The patient subsequently underwent complete surgical excision of the posterior mediastinal mass, without postoperative complications.

Follow-up chest radiography and thoracic CT scan demonstrated complete resolution of the mass, with satisfactory lung re-expansion. A small ipsilateral pleural effusion persisted, along with postoperative changes and a chest drain in situ (Figure 3-4).

Histopathological examination of the surgical specimen confirmed a grade 1 immature pluritissular teratoma.

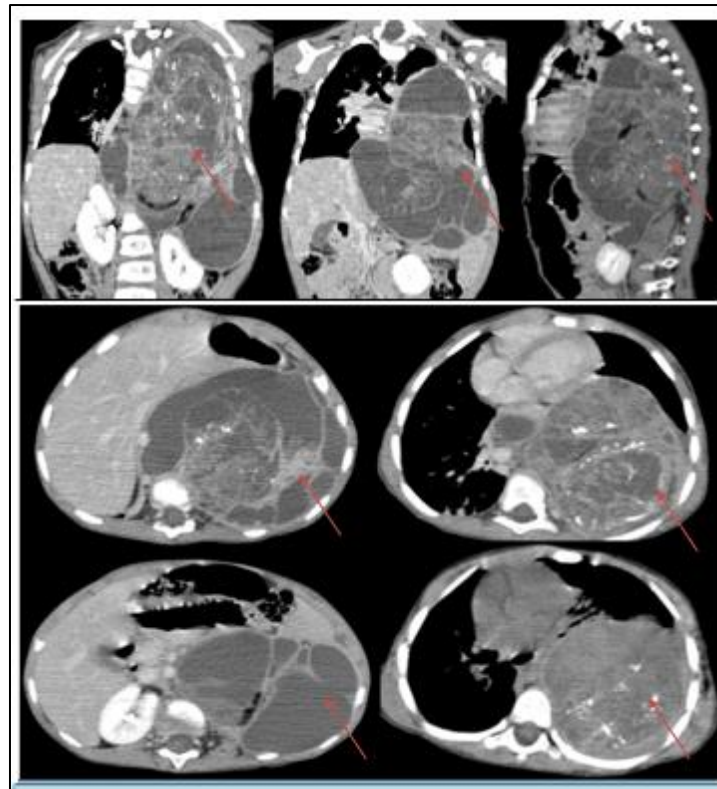
The postoperative course was uneventful, with favorable clinical and radiological outcomes. The patient was placed under regular active surveillance, which remains satisfactory to date.

A follow-up thoraco-abdominal CT scan performed two years after surgery showed complete resolution of the mass, with no residual lesion or recurrence (Figure 5).



Reference: Radiology department CHU HASSAN II, FEZ, Morocco

Figure 1 Frontal chest radiograph demonstrates a heterogeneous fluid-density opacity involving the left hemithorax, without obscuration of the cardiac silhouette and without air bronchograms



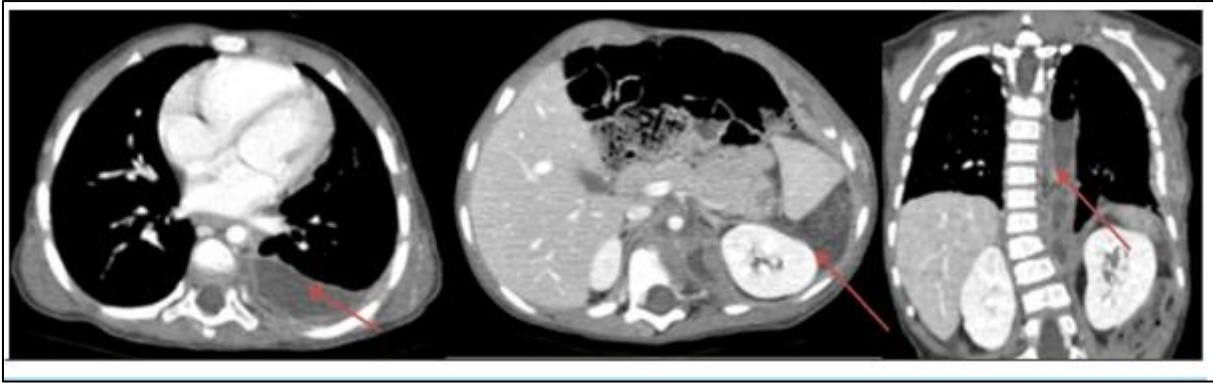
Reference: Radiology department CHU HASSAN II, FEZ, Morocco

Figure 2 Thoracoabdominal CT scan with axial and coronal reconstructions revealed a large, poorly defined left posterior mediastinal mass occupying nearly the entire left hemithorax. The lesion was mixed solid and cystic, containing scattered calcifications and fatty components, measuring approximately 9.5 × 10 × 18 cm (anteroposterior × transverse × craniocaudal). It exerted a significant mass effect, displacing the descending aorta, inferior vena cava, azygos vein, and tracheobronchial tree to the right, as well as the diaphragm, left kidney, spleen, and transverse colon inferiorly



Reference: Radiology department CHU HASSAN II, FEZ, Morocco

Figure 3 The postoperative frontal chest radiograph showed complete resolution of the mass, with satisfactory lung re-expansion and a chest drain in place



Reference: Radiology department CHU HASSAN II, FEZ, Morocco

Figure 4 Postoperative thoracic CT scan with axial and coronal reconstructions demonstrates complete resolution of the mass, with satisfactory lung re-expansion and persistence of a small ipsilateral pleural effusion



Reference: Radiology department CHU HASSAN II, FEZ, Morocco

Figure 5 Thoracoabdominal CT scan performed two years after surgery demonstrated complete resolution of the mass, with no residual lesion or evidence of recurrence

3. Discussion

The term “teratoma” derives from the Greek word *teras*, meaning “monster” [6]. Teratomas originate from the three embryonic germ layers and are classified as mature (benign) when their tissues are well differentiated, or immature (malignant) when they contain predominantly undifferentiated elements [7].

In some cases, both mature and immature components coexist within the same tumor; these lesions are referred to as mixed teratomas, reflecting a wide spectrum of histological combinations.

Teratomas can arise in the gonads, along the midline structures of the central nervous system, in the sacrococcygeal region in children, and within the mediastinum [4]. Mature mediastinal teratomas account for approximately 8% of mediastinal tumors and 44% of germ cell tumors, with no clear sex predilection [4,5].

Isolated involvement of the posterior or middle mediastinum is uncommon, occurring in approximately 2–8% of cases [1].

The pathogenesis of extragonadal germ cell tumors remains incompletely understood. The most widely accepted hypothesis is an arrest in the migration of primordial germ cells to the genital ridges during embryogenesis [8]. An alternative theory suggests a reverse migration of germ cells from the gonads toward extragonadal sites [9].

Clinically, the majority of patients remain asymptomatic, with mediastinal masses most often identified incidentally during thoracic imaging performed for unrelated indications [1].

Masses typically become symptomatic due to compression of mediastinal structures, which may lead to respiratory distress, particularly in infants, respiratory insufficiency, superior mediastinal syndrome, cervical mass, or Claude Bernard–Horner syndrome. They may also present through ectopic hormonal secretion, such as β -hCG or insulin [1].

Tumor rupture into the thoracic cavity may result in acute clinical manifestations, including chest pain, hemoptysis, respiratory distress, cardiac tamponade, or pleural effusion [1].

In rare cases, trichoptysis may be observed, indicating a communication between the tumor and the airways [10].

Chest radiography may reveal a well-defined anterior mediastinal opacity, with calcifications observed in approximately 26% of cases of mature teratomas [11]. The presence of teeth or well-formed osseous elements is rare but highly suggestive of the diagnosis.

Computed tomography (CT) is the imaging modality of choice, as it accurately localizes the mass and characterizes its various components, while also assessing its relationship with adjacent structures.

Magnetic resonance imaging (MRI) is particularly useful for evaluating possible invasion of surrounding tissues or in complex thoracic locations, such as the thoracic inlet or thoracoabdominal extension [12].

Assessment of tumor markers, including α -fetoprotein (α FP) for yolk sac tumors and β -hCG for choriocarcinoma, is essential when an immature component is suspected or in the workup of other germ cell tumors [3].

On chest radiography, the differential diagnosis of mediastinal masses is broad and includes all possible etiologies arising in this location [1].

However on computed tomography (CT), the diagnostic spectrum becomes narrower, and interpretation is mainly guided by the lesion's internal composition. When a fatty component is present, thymolipoma should be considered. Soft-tissue masses, often heterogeneous, suggest germ cell tumors, lymphoma, or thymic neoplasms. When the lesion is predominantly cystic, the main differential diagnoses include thymic cyst and bronchogenic cyst [1].

Complete surgical resection of mature mediastinal teratomas is usually curative. However, when an immature component is present, adjuvant chemotherapy is required. Overall, the prognosis of mature teratomas is excellent, although thorough histopathological evaluation is essential to rule out any immature elements [2].

4. Conclusion

Germ cell tumors, particularly teratomas, should be considered in the differential diagnosis of any mediastinal mass in children and young adults. Imaging plays a key role in diagnosis, lesion characterization, and staging. Management is based on complete surgical resection, complemented by thorough histopathological evaluation to identify any immature components. This approach helps optimize prognosis and guide further management.

List of abbreviations:

- **CT** : Computed tomography
- **AFP** : Alpha-fetoprotein
- **β -hCG** : Beta-human chorionic gonadotropin
- **MRI** : Magnetic resonance imaging

Compliance with ethical standards

Disclosure of conflict of interest

The authors do not declare any conflict of interest

Availability of data and materials

The data sets are generated on the data system of the CHU Hassan II of Fes, including the biological and radiological data.

Statement of informed consent

Written informed consent was obtained from the patient. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Author's contribution

- KL Is the corresponding author, she participated in the organization and writing of the article.
- Professor MB supervised the working and validated the figures.
- Professor and chief of department of radiology MB and MM read and allowed the article for publication.
- All authors read and approved the final manuscript.

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