Pleural effusion revealing a malignant rhabdoid tumor of the chest wall in an infant: A case report and literature review

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ABSTRACT

Introduction: The rhabdoid tumor (RT) is a malignant tumor, rare and aggressive, with a poor prognosis. It affects the young children, especially the infants. Described for the first time in 1978 by Beckwith and Palmer, it develops mainly in the kidney but other localizations like cerebral and extra-cerebral have been described. The mediastinal and pulmonary localizations remain rare. Case Report: We report a case of RT of an 18-month-old male infant admitted for pleural effusion syndrome, without any past medical history. The chest X-ray revealed a large opacity of the left hemithorax compressing heart and the mediastinum structures to the right. It was associated with a lysis of the sixth rib. Thoracic computed tomography (CT*) showed a large lobulated heterogeneously parietal mass delimiting liquid hypodense area with pleural effusion. The mass compressed the homolateral lung with reduction of left lobar bronchus and mediastinal structures to the right. It was associated with a lysis of the fifth and sixth ribs with soft tissue extension. Histopathological examination revealed richly vascularized fibrous tissue composed of large cell ranges with reduced cytoplasm and oval nuclei with vesicular chromatin, and cell ranges with eccentric nuclei and deep eosinophilic cytoplasmic inclusion. On immunohistochemistry, integrase interactor 1 (INI-1) protein labeling was negative. The diagnosis of the RT was retained. A chemotherapy was started, but despite chemotherapy the patient's condition was deteriorated on the respiratory with dependence on oxygen therapy. The patient died after 2 months and 10 days of diagnosis. Conclusion: The RT is a rare entity occurring usually in young children very frequently in infants. It develops mainly in the kidney but other localizations have been described including the mediastinal and pulmonary localizations which are very rare. It is extremely aggressive in nature and quickly progresses. The imaging does not provide pathognomonic diagnostic signs. The histopathological and immunohistochemical study with immunolabeling makes it possible to clarify the diagnosis by the absence of labeling of the protein integrase interactor. Generally, the prognosis remains very poor despite the chemotherapy and radiotherapy until now.

Keywords: Chest wall, Extra-renal tumor, INI-1, Malignant rhabdoid tumor

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INTRODUCTION

The rhabdoid tumor is a malignant tumor, rare and aggressive, with a poor prognosis. It affects the young children, especially the infants [1, 2]. Described for the first time in 1978 by Beckwith and Palmer, it develops mainly in the kidney but other localizations like cerebral and extra-cerebral have been described [3–8]. The tumor of central nervous system was called the atypical teratoid/rhabdoid tumors (AT/RTs). The mediastinal and pulmonary localizations remain rare [5, 6]. Here, we describe a case of malignant RT of the chest wall in an infant.

CASE REPORT

We report the case of an 18-month-old male infant, without past medicals history. The beginning of the symptomatology returned to three weeks of its admission by a dry cough, polypnea, and fever without improvement despite empiric antibiotic therapy. The chest radiography was practiced and showed a left pleural effusion with lysis of sixth rib and the patient was admitted in the pediatric unit.

On admission the patient was in good general condition, reactive, with cutaneo-mucous pallor, and respiratory distress (polypnea, decrease vesicular murmur of the lower 2/3 of the left lung), however he was hemodynamically stable. The patient had a left thoracic parietal swelling adhering to the wall estimated at 4 cm. The rest of the exam was featureless.

Biological: hemoglobin was at 9.4 g/dL, leukocyte at 18,140/mm, renal function and ionogram were normal.

In imaging, the chest X-ray revealed a large opacity of the left hemithorax compressing heart and the mediastinum structures to the right. It was associated with a lysis of the sixth rib (Figure 1).

Thoracic computed tomography (CT*) showed a large lobulated heterogeneously parietal mass delimiting liquid hypodense areas with pleural effusion measuring 90 × 67 × 86 mm. The mass compressed the homolateral lung with reduction of left lobar bronchus and mediastinal structures to the right. It was associated with a lysis of the fifth and sixth ribs with soft tissue extension (Figure 2).

The histological samples were taken on the left thoracic parietal swelling, the site of the tumoral extension. Histopathological examination revealed richly vascularized fibrous tissue composed of large cell ranges with reduced cytoplasm and oval nuclei with vesicular chromatin, and cell ranges with nuclei often eccentric with abundant eosinophilic cytoplasmic inclusion concluding to a malignant tumor process difficult to classify (Figure 3).

On immunohistochemistry, cytokeratin staining, vimentin, epithelial membrane antigen, smooth muscle actin, and CD99 were positive. Labeling with LCA-CD3–CD20-ALK-CD30, MPO-CD34-CD68, CD1, desmin–myogenin were negative. Also, INI-1 protein labeling was negative.

With the quickly evolving clinical picture and the histopathological result in particular the absence of labeling of the protein INI-1, the diagnosis of the RT was retained two weeks after his hospitalization.

Figure 1: Opacity of the left hemithorax compressing heart and the mediastinum structures to the right. It was associated with a lysis of the sixth rib.

Figure 2: (A) Axial CT without injection showing a discretely hyperdense left parietal process compressed homolateral lung and mediastinum structures to the right with bone lysis. (B) and (C) Axial and coronal CT with injection showing a heterogeneous mass delimiting liquid hypodense areas with pleural effusion. The mass compressed the homolateral lung with reduction of left lobar bronchus and mediastinal structures to the right. It was associated with a lysis of the fifth and sixth ribs with soft tissue extension. (D) 3D bone reconstruction showing bone lysis of the sixth left rib.
A chemotherapy treatment was started with associated vincristine, actinomycin, cyclophosphamide, then with cyclophosphamide/Adria and VP16/carboplatin alternately.

Despite chemotherapy the patient’s conditions was deteriorated on the respiratory with dependence on oxygen therapy. He was also suffering from severe anemia which it was transfused by a red blood cell. The patient was died after 2 months and 10 days of diagnosis.

**DISCUSSION**

The RT was described for the first time in 1978 by Beckwith and Palmer as a “rhabdomyosarcomatoid variant of Wilms tumor” with high grade of malignancy and affecting the infant [3]. It is a rare tumor, extremely aggressive with poor prognosis [1, 2]. It usually occurs in young children very frequently in infants under two years old. The median age at the time of diagnosis is ranging from 10 to 24 months depending on the series [9].

The incidence is 5 per 1,000,000 in the first year of life and decreased by age to 0, 6 per 1,000,000 at age 1–4 years, 0, 1 per 1,000,000 at age 5–9 years. The sex ratio is equal at 1, 08/1, however AT/RT predominated in boys [10, 11].

This tumor develops usually in the kidney [3]. However, extra renal localizations such as the nervous system, liver, and soft tissues of digestive tract, lung, and mediastinum have been described [3–8, 12]. The tumor of central nervous system was called the AT/RT. About less than 34 cases of mediastinal localization have been reported to date, from which 9 cases are related to children less than 18 years [6]. Less than 40 cases of pulmonary primitive localization of all ages were reported [5, 13, 14].

Clinically, the mode of revelation is very polymorphic and depends on the localization of the tumor. The RT of kidney revealed by hematuria in 80% of cases and associated with hypocalcemia in 20% of cases. In the nervous system tumor is revealed by intracranial hypertension or cerebellar syndrome and in the thoracic level it manifested by chest pain, dyspnea, cough, or respiratory distress [6, 15–17].

The imaging does not provide pathognomonic diagnostic signs. Some authors describe the heterogeneous nature of the lesion with intense contrast enhancement often multilobulated with some areas of necrosis or intra-tumor calcification [6, 14, 18, 19].

Anatomo-pathological examination is very variable, often marked by the presence of rhabdoid cells, composed of a nucleus with a large single nucleolus and a decompacted chromatin seat of an intracytoplasmic eosinophilic inclusion, within an undifferentiated stroma making the diagnosis difficult [14, 20, 21]. The immunohistochemical study with immunolabeling eliminates the differential diagnosis and suggests the diagnosis of the RT. Indeed, the absence of labeling of the protein INI-1 which is the result of the loss of the function of the suppressor gene SMARCB1 makes it possible to specify the diagnosis [14, 20–22].

The treatment of RT is based on radical surgery. Hilden et al. have been reported that the patients who underwent radical surgery had a higher survival than those who underwent partial surgery. But this radical surgery could not be realized because at the time of diagnosis the tumor was too large [23, 24]. Some authors have been reported the beneficial role of chemotherapy and radiotherapy [25–27]. There is no consensual therapeutic protocol for chemotherapy or radiotherapy. Some authors and European Commission recommend intensify (high dose) chemotherapy and radiotherapy over 18 years and the results seem promising [26, 28, 29].

Despite all these treatments, the prognosis of these tumors remains very poor. The average survival varies according to studies from 11 to 17 months [1, 2, 6].

**CONCLUSION**

The RT is a rare entity occurring usually in young children very frequently in infants. It develops mainly in the kidney but other localizations have been described including the mediastinal and pulmonary localizations which are very rare. It is extremely aggressive in nature and quickly progresses. The imaging does not provide pathognomonic diagnostic signs. The histopathological and immunohistochemical study with immunolabeling makes it possible to clarify the diagnosis by the absence of labeling of the protein INI-1 which is the result of the loss of the function of the suppressor gene SMARCB1 makes it possible to specify the diagnosis [14, 20–22].
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intellectual content, Final approval of the version to be published, Agree to be 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

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Authors declare no conflict of interest.

Data Availability
All relevant data are within the paper and its Supporting Information files.

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