Case Report Primary pulmonary spindle cell rhabdomyosarcoma in adolescent: a case report and review of literatures

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Abstract: Spindle cell rhabdomyosarcoma (SRMS) is a rare and new type of rhabdomyosarcoma. SRMS in lung is extremely rarely. Here we report on a first explicit case of SRMS in lungs of a 13-year-old girl. Macroscopic examination showed a tumor (65 mm×17 mm×9 mm) with a solid gray-white cut surface which displays invasive growth but was well-circumscribed. Histologically, tumor cells were formed with spindle to round cells with eosinophilic cytoplasm and fascicular, storiform, wavy appearance. Horizontal grain structure and striated muscle brood cells were observed in some cells or local regions. A small amount of collagen fibers were observed in the mesenchyme. The nucleus was long or wavy with nuclear heteromorphosis. Immunohistochemically, tumor cells were stained diffusely positive for muscle specific actin, desmin, and vimentin, scattered positive for myogenin, MyoD1 and myoglobin. According to the examination results, this case was diagnosed as SRMS. Then the tumor was completely excised with a part of lung tissues, and additional chemotherapy was given.

Keywords: Rhabdomyosarcoma, spindle cell, diagnoses

Introduction

Rhabdomyosarcoma (RMS) is the most common soft-tissue sarcoma in children under 15 years of age [1]. Spindle cell rhabdomyosrcoma (SRMS) is a rare and new type of RMS. It usually occurs among children and adolescents paratesticular, followed by head and neck section, with a better prognosis in comparison with other subtypes of RMS. In 1995, the International Classification of RMS had provided prognostically classification for RMS and allowed risk stratification for children with RMS. There are three major histologic variations of this tumor: embryonal RMS (ERMS), alveolar RMS (ARMS) and pleomorphic RMS (PRMS). Thereinto ERMS is divided into the conventional, botryoid, spindle cell/sclerosing, and anaplastic variant. As both spindle cell and sclerosing RMS have similar clinical presentations, some scholars suggested that they may represent a histologic spectrum of a single pathologic [2]. More recently, the 2013 WHO reclassified spindle cell/Sclerosing as a stand-alone pathologic entity, separate from embryonal RMS [3].

Primary pulmonary neoplasms are unusual in the pediatric age group and RMS of the lung is one of the rarest malignancies. To our knowledge, only a few pediatric cases have been reported in the literature [4]. It accounts only 0.5% of childhood RMS [5]. SRMS in lungs is extremely uncommon. Then as an independent subtype of RMS, limited reported cases, unclear clinical characteristics and genetic changes, insufficient understanding can easily lead to misdiagnosis and undiagnosis. Therefore it is of great significance for us to report the case a 13-year-old girl who had primary SRMS in lung together with a discussion on the differential diagnosis of this lesion.

Case presentation

A 13 young girl presented with cough company right chest pain and fever was admitted to our



Figure 1. Spindle cell RMS. (A, B) The tumor cells were formed with spindle to round cells with eosinophilic cytoplasm and fascicular, storiform, wavy appearance. A small amount of collagen fibers were observed in the mesenchyme. The nucleus was long or wavy with unsignificantly nuclear heteromorphosis. (C) Striated muscle brood cells were visible. (D) Horizontal grain structure and mitotic (2-5/10 high power filed) were observed in some cells or local regions. H&E, original magnification: (A) ×100; (B, C) ×200; (D) ×400.

hospital. Laboratory tests indicated no significant abnormality. Chest X-ray indicated a mass 70 mm in diameter with focal solid change in lower lobe of right lung. Complete and extended surgical of inferior lobe of right lung. Macroscopic examination showed a tumor (65 mm×17 mm×9 mm) with a solid gray-white cut surface which displayed invasive growth but were well-circumscribed. Histologically, tumor cells were formed with spindle to round cells with eosinophilic cytoplasm and fascicular, storiform, wavy appearance. A small amount of collagen fibers were observed in the mesenchyme. The nucleus was long or wavy with nuclear heteromorphosis (Figure 1A and 1B). Striated muscle brood cells were observed. Horizontal grain structure and mitotic were visible in some cells or local regions (Figure 1C and **1D**).

Immunohistochemistry, the tumor cells were stained positively for muscle-specific actin,

desmin (Figure 2C), vimentin and myoglobin. Meanwhile, tumor cells also showed positively for myogenin (Figure 2A) and myoD1 (Figure 2B). S100 (Figure 2D) and NSE (Neuron Specific Enolase) were not present. Furthermore the molecular genetic testing to check for the presence of Pax3-FOX01 and Pax7-FOX01 fusion gene was also negative.

Based on histopathologic and immunohistochemical findings, the tumor was diagnosed as a SRMS. The tumor was completely excised with a part of lung tissues, and additional chemotherapy was given.

Discussion

RMS originates from the embryonal mesenchyme that ultimately gives rise to striated skeletal muscle. It develops mostly in the head and neck, genitourinary tract, and retroperitoneum



Figure 2. Immunohistochemistry of spindle cell RMS. The tumor cells were positively stained for myogenin (A), MyoD1 (B). Scattered tumor cells had strongly positive for desmin (C). S100 was absolutely negative (D). (A-D) ×200.

[6]. SRMS is a new rare subtype of RMS mainly seen in children and accounting for 4.4% of all RMS subtypes [7]. This was reported firstly by Cavazzana et al in 1992 [8]. Some scholars found that there is sclerosing RMS area in SRMS, and vice versa. Although the two are closely linked, in most cases are given priority to a form. Herein we present a case as spindle cell histologic type of adolescence in pulmonary.

SRMS in lung is an extremely rare malignancy occuring in children and adults on account of the fact that the lung is lack of striated muscle. The Intergroup RMS Study (IRS) Committee conducted a study on thoracic sarcoma in children. They revealed a total of 84 patients presenting with thoracic sarcoma. Of these, only 3 were lung [9]. Primary pulmonary RMS always occur as embryonal or alveolar variants in children or as the pleomorphic variant in adults [10]. As a special site, the definitive diagnosis methods of SRMS in lung are in accord with SRMS in other sites. The pathological changes of SRMS are mainly as follows. Histological examination showed a highly cellular tumor composed of spindle cells with elongated, vesicular nuclei, inconspicuous nucleoli and eosinophilic cytoplasm, arranged in a herringbone pattern which is similar with rare fibrosarcoma in adults, leiomyosarcoma or malignant peripheral nerve sheath tumor [11]. Horizontal grain structure and striated muscle brood cells were observed in some cells or local regions like contractile fiber cells. Mitotic is visible, generally from 0 to 7/10 high power filed. The spindle cell morphology in the tumor presented here initially provided some diagnostic challenges, thus an immunohistochemical staining using a panel of markers was necessary. Some markers of RMS have been used to detect myoid differentiation, such as desmin, actin and myoglobin. These markers are always associated with the degree of tumor cell differentiation. Recent study found that MyoD1 and myogenin are considered sensitive and specific markers for SRMS and are more specific than desmin and muscle-specific actin and more

sensitive than myoglobin [12]. In our case, the spindle tumor cells are uniformly and strongly positive for all muscle cell markers (actin, myoglobin, desmin, vimentin), and also positive for myogenin and MyoD1 in spindled tumor cells in addition to obvious rhabdomyoblasts, keeping with immunophenotypic features of an embryonal RMS. Thus, the diagnosis of SRMS was preferred.

Primary pulmonary SRMS is rare and can be easily misdiagnosed. It should be differentiated from leiomyosarcoma, mixed epithelial, mesenchymal tumors and some other small cell lung cancers.

SRMS has a good prognosis. The clinical biological behavior has difference between SRMS in adults and in children. Generally the prognosis of children is better than adults [8]. SRMS is treated with similar protocols as for other RMS. The therapeutic schedule involves combined therapy including surgery, chemotherapy, and adjuvant radiation.

In summary, primary pulmonary SRMS is rare and can be easily misdiagnosed. Reporting a large series of these tumors may lead to a better understanding aid in a diagnosis of pulmonary SRMS.

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Disclosure of conflict of interest

None.

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