CASE REPORT

EXPERIMENTAL ONCOLOGY

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AGGRESSIVE RHABDOID TUMOR IN THE RIGHT LUNG: A CASE REPORT

Rhabdoid tumor is a rare and aggressive neoplasm that usually occurs in children and is often localized in the central nervous system and kidneys, but can be found in many other sites. In our case report, we describe a tumor that was found on computed tomography in the thoracic region of a 62-year-old male and was successfully surgically resected. The images and descriptions of our findings and the results of the additional immunohistochemical studies allow us to make the final diagnosis.

Keywords: rhabdoid tumor, immunohistochemistry, diagnosis.

Rhabdoid tumor (RT) is an aggressive neoplasm that can occur in many anatomic sites, including the kidneys, central nervous system, axial locations such as the neck, paraspinal region, perineal region, abdominal cavity or retroperitoneum, pelvic cavity, skin, and other localizations [1]. RT may consist of rhabdoid cells only while in some cases, a combination of primitive neuroectodermal cells, mesenchymal cells, and/or epithelial cells is observed [2]. According to the WHO Classification of Tumours of Soft Tissue and Bone Tumours, the 5th revision, RT belongs to soft tissue tumors/tumors of uncertain differentiation and have ICD-O code: 8963/3, rhabdoid tumor, NOS [3].

Clinically, the tumor usually is presented as a rapidly growing soft tissue mass, whose clinical manifestations depend on its localization. The tumor can be localized in different organs, in particular in the omentum, possessing different growth patterns [4]. Multiple cutaneous nodules may be seen [5], and in pediatric cases, there may be a disseminated disease without any primary tumor [6]. The localization of the tumor in the kidneys or brain is characteristic of children and adolescents, while tumors outside these two localizations are characteristic of the older age groups [7].

The etiology of RT is still being studied. The association of the familial cases with the germline

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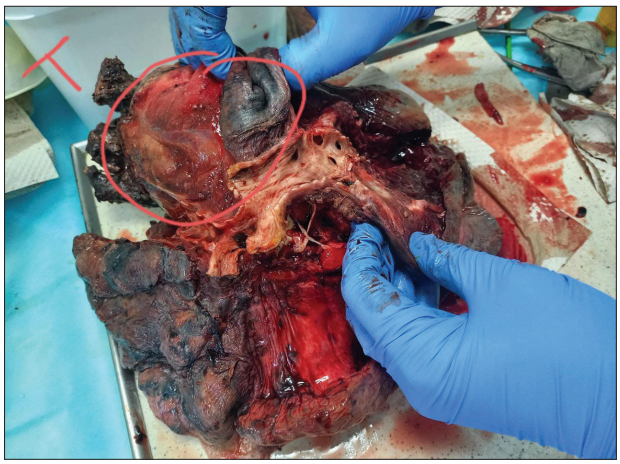


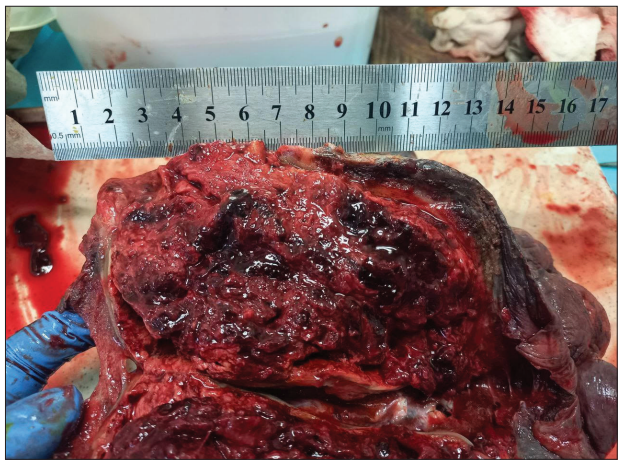
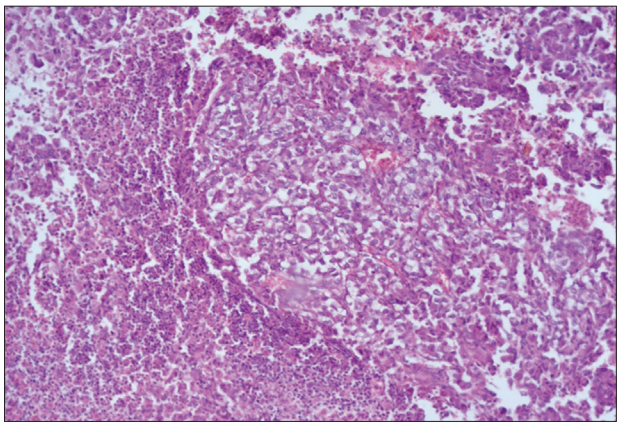


Fig. 1. Resected postoperative material of the patient with a tumor (T) growing into the surrounding soft tissues and ribs

Fig. 2. Macroscopic view of tumor on cut-section showing brownish-reddish mass lesion up to 11 cm with a whitish-yellow capsule



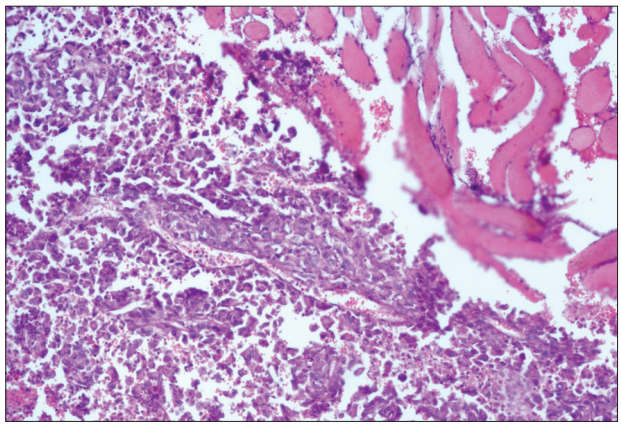


Fig. 3. Rhabdoid cells with eosinophilic cytoplasm, vesicular eccentrically located nuclei with nucleoli. Hematoxylin and eosin staining, x100

Fig. 4. Rhabdoid cells that grow into the surrounding muscle tissue. Hematoxylin and eosin staining, ×100

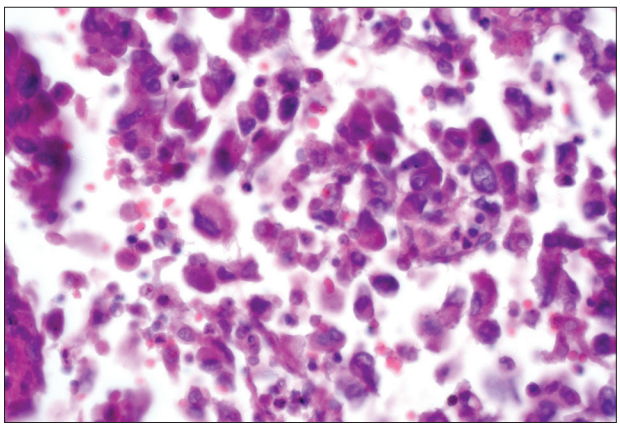
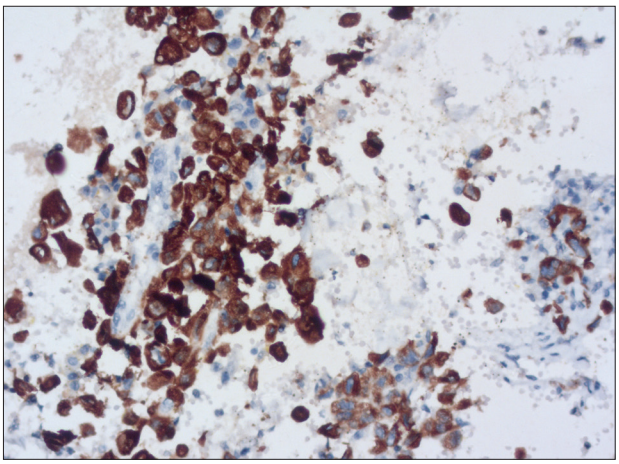


Fig. 5. Rhabdoid cells with eosinophilic cytoplasm and nucleoli demonstrating discohesive growth. Hematoxylin and eosin staining, ×400

mutations in *SMARCB1* (INI1, hSNF5, BAF47) in 22q11.23 has been proved [6]. The germline mutations or deletions in *SMARCB1* (RT predisposition syndrome 1) are seen in approximately 13% of patients with extrarenal RTs [8].

The pathogenesis of RT consists in the homozygous inactivation of the *SMARCB1* gene in 22q11.2. Approximately 98% of extrarenal RT, malignant renal RT, and atypical teratoid/rhabdoid tumors show the genomic alterations of both copies of the gene, including coding sequence mutations, partial or whole gene deletions, and a loss of the neutral copy. As a result of these mutations, in the state of heterozygosity, a recessive allele is manifested on the remaining homolog [9].



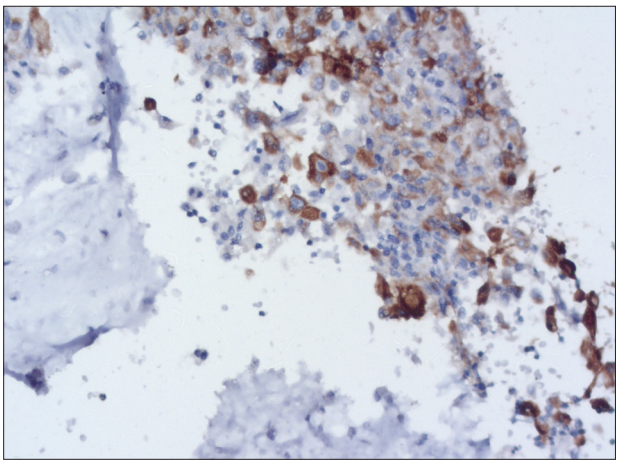


Fig. 6. Tumor cells were positive for AE1/AE3. IHC staining for AE1/AE3 (AB-1 clone), ×200

Fig. 7. Tumor cells were positive for CK7. IHC staining for CK7 (AB-2 clone), ×200

The differential diagnosis of RT can be complicated as it should be differentiated with:

- epithelioid sarcoma, where one can observe the nodular proliferation of epithelioid cells with the focal rhabdoid features and positive immunohistochemical (IHC) staining on vimentin, cytokeratin, EMA, and often CD34 with a loss of INI-1 [10, 11];
- rhabdomyosarcoma, which is characterized by positive muscle markers and positive INI-1 expression [12];
- desmoplastic small round cell tumor with desmoplastic stroma and positive desmin staining, also with positive INI-1 expression [13];
- epithelioid malignant peripheral nerve sheath tumor, where we can observe relatively uniform atypical epithelioid cells with diffuse staining for S100 and loss of INI-1 expression [11—13].

In the current publication, we present a case of a 62-year-old male who was treated in the Department of Thoracic Oncosurgery at the Clinical Hospital "Feofaniya" (Kyiv, Ukraine) and initially presented with a mass lesion on computed tomography (CT) in the upper parts of the right lung. Extrapulmonary mass had a cystic-solid structure with clear contours, and the total dimensions of this mass were 11.8 × 7.6 × 8.2 cm. The tumor originated from the costal pleura, adjacent to the 6th and 7th ribs with a minor remodeling of their internal contour, and in the 6th rib, a consolidating fracture was found. The lumen of the trachea and main bronchi were not changed. Intrathoracic lymph nodes were not enlarged. The mediastinum

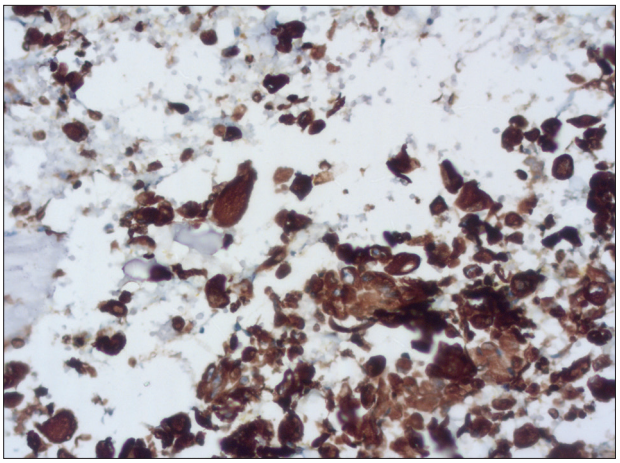


Fig. 8. Tumor cells were positive for vimentin. IHC staining for vimentin (SP20 clone), ×200

was not displaced. The data of the CT study were considered by the radiologist as "CT signs of a mass lesion in the upper parts of the right hemithorax, originating from the costal pleura and adjacent to the 6th and 7th ribs with their minor remodeling; the 6th rib at this level with a consolidating fracture".

A surgical treatment was performed — excision of ribs 5—7 and pneumonectomy. The images of the resected operative material are presented in Fig. 1.

The grossing findings of the post-operative specimen were as follows: the tumor was up to 11 cm in size, originating from the costal pleura and adjacent to the ribs with a consolidating fracture in one of them, surrounded by a whitish-yellow capsule, brownish-reddish in cross-section, with a lobular structure (Fig. 2).

The microscopic examination revealed rhabdoid cells with eosinophilic cytoplasm, vesicular eccentrically located nuclei with nucleoli, sprouting into surrounding tissues (Figs. 3, 4).

Tumor cells were polymorphic, and on ×400 magnification, some of them were large with eosinophilic cytoplasm and an increase in the number of nucleoli in the nucleus; there were observed pathological mitoses (Fig. 5).

The initial pathomorphological analysis of the specimen and the clinical data suggested a diagnosis of high-grade carcinoma of squamous histogenesis. We need to exclude a whole range of tumors of anaplastic morphology including melanoma.

The IHC study was performed to verify the diagnosis. We used the monoclonal antibodies mentioned below manufactured by Master Diagnostica (Brasil). Positive expressions of AE1/AE3, SK7, and vimentin were found in tumor cells (Figs. 6—8).

The second pathology conclusion was a malignant dedifferentiated neoplasm of the right lung.

At the final stage of the diagnostic process, an analysis of the morphological and IHC findings with additional IHC markers showed that the tumor did not exhibit differentiation markers from mesothelium, melanin-forming, germinogenic, vascular, and smooth and striated muscle tissues. We noted that some tumor cells, including polymorphic large cells of the rhabdoid type, express CK7, AE1/AE3, and vimentin, and IHC reactions with monoclonal antibodies to CK5/6, CK20, CD34, SMA, napsin A, TTF1, calretinin, and WT1 were negative, which cannot exclude epithelial histogenesis and/or certain types of sarcoma.

After analyzing the obtained histological, IHC data (positive expression of CK7, AE1/AE3, and negative IHC reactions with monoclonal antibodies to CK5/6, CK20, CD34, SMA, napsin A, TTF1, calretinin, and WT1) and CT report, taking into account the latest data from literature sources [3, 14—16], we can conclude that the patient had a rhabdoid tumor of soft tissues/malignant rhabdoid tumor with invasion into the costal pleura with a transition to adjacent ribs 6-7 and involvement of the perifocal soft tissues (ICD-O: 8963/3 — rhabdoid tumor, NOS).

Diagnosis of rhabdoid tumors is difficult and requires a transdisciplinary approach with the use of a wide range of additional methods using a large number of IHC markers and, if possible, molecular genetic studies.

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АГРЕСИВНА РАБДОЇДНА ПУХЛИНА ПРАВОЇ ЛЕГЕНІ: КЛІНІЧНИЙ ВИПАДОК

Рабдоїдна пухлина — це рідкісне та агресивне новоутворення, яке зазвичай виникає у дітей і часто локалізується в центральній нервовій системі та нирках, але може бути виявлена в багатьох інших місцях. Ми описуємо пухлину, виявлену в грудній ділянці 62-річного чоловіка на комп'ютерній томографії та успішно видалену хірургічним шляхом. Надано зображення та описи наших патогістологічних знахідок та дані додаткових імуногістохімічних досліджень, що дозволило нам поставити остаточний діагноз.

Ключові слова: рабдоїдна пухлина, імуногістохімічне дослідження, діагноз.