Pathomorphological features of rhabdomyosarcomas in pediatric patients

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Objective. We want to explore the specific morphological manifestations, determine the frequency of different histological variants, as well as identify local mutations of genes determining the development of these tumors.

Methods. Medical records and histological preparations of 7 children treated with the diagnosis were analyzed at the pathologoanatomic department of St. Petersburg State Medical University: "Rhabdomyosarcoma" from 2001 to 2018. Histological preparations were photographed using a Pannoramic Midi 2 scanning microscope.

Results.

- Numerous star-shaped and elongated, spindle-shaped cells irregularly distributed among the stroma were found in embryonic RMS (ERMS) preparations. Elongated cells with eosinophilic cytoplasm with transverse striations in the cytoplasm were also detected.
- In pleomorphic RMS, tumor cells were determined to have a variety of shapes, including rocket-shaped, with oval nuclei and small nuclei. Large foci of myxomatosis of tumor stroma were observed.
- Alveolar rhabdomyosarcoma (ARMS) was characterized by the presence or formation of alveolar structures lined by rounded or oval-shaped tumor cells. The nuclei of these cells were kidney-shaped and/or lobulated, with a well-defined cytoplasm. Hyalinized fibrous septa were detected.
- Alveolar and pleomorphic RMS are formed from elements of muscle tissue, and in rare cases alveolar from endothelial progenitor cells after reprogramming and myogenic transdifferentiation, while ERMS has a dysontogenic origin, that is arises from detached rudiments of muscle tissue and are transverse muscular tissue hamartoblastomas.
- Alveolar RMS is a more malignant and less differentiated tumor, for which reason the mortality rate in patients with ARMS is significantly higher than in other types of RMS.
- ARMS are associated with the *FOXO1* gene fusion return system found in 90% of cases.
- 56 mutations were found in 28% of ERMS cases and included 7 mutations in rate in the RAS family, 4 mutations in *FGFR4*, 3 mutations in *PIK3CA*, 2 mutations in *CTNNB1* and single mutations in *BRAF* and *PTPN11*.