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abstract

DICER1 Mutant Sarcomas with Rhabdomyosarcoma-Like Features: Pathological and epidemiological diversity of cases in single reference center

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abstract

DICER1 Mutant Sarcomas with Rhabdomyosarcoma-Like Features: Pathological and epidemiological diversity of cases in single reference center

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Introduction: DICER1 mutant sarcoma is a rare entity within tumors with rhabdomyosarcoma-like features. It seems to be more aggressive than rhabdomyosarcomas and has poor outcome according to literature. The purpose of this study was to analyze patients with these tumors diagnosed in one reference center.

Methodology: During the period of 2017-2024 (96 months) 7 patients with different localizations were diagnosed as DICER1-mutant sarcomas due to morphology and NGS. All patients received chemotherapy after biopsy according to CWS protocol.

Results: Age of patients varied from 26 to 173 months (two patients under 4 years and five patients above 12 years). Localizations of tumors were fallopian tube, pelvis (2), cervix, vagina, larynx, abdomen. Biopsies were performed in all cases before chemotherapy. Morphological study showed spindle cell morphology with irregular cellularity and myogenic immunophenotype. Two cases showed cartilage within the lesions. Only in two cases DICER1-sarcoma was suspected, other diagnoses were embryonal rhabdomyosarcoma (1), undifferentiated sarcoma (1), spindle cell rhabdomyosarcoma (1), sarcoma with SMARCA4 rearrangement (1), teratoma with somatic type of malignancy (1). NGS showed various types of DICER1 mutations. All patients received or still receive chemotherapy according to CWS protocol. Median follow- up time was 36 months (min. 7 months, max. 86 months). 6 patients are alive with no progression (no information about 1 patient).

Conclusion: The presented data shows variable histology and equal immunophenotype in DICER1 mutant sarcomas that should be considered in differential diagnosis in cases with spindle cell myogenic morphology.