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abstract

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The malignant tumour spectrum in children with neurofibromatosis type 1

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Introduction: Neurofibromatosis type 1 (NF1) is one of the most common cancer predisposition syndromes. Mainly, patients with NF1 have benign tumours, primarily, plexiform neurofibromas. However, some of them develop malignant tumours and require special treatment such as chemotherapy, radiation or target therapy.

Methodology: The cohort includes 8 patients with genetically confirmed NF1 who were diagnosed with malignant tumour. There were 6 girls and 2 boys, the median age was 12 years old (4-17 y.o.). All children were observed in Dmitry Rogachev National Medical Research Center of Pediatric Hematology, Oncology and Immunology in 2021-2024. All patients received special treatment according to the tumour type.

Results: The tumour spectrum includes Malignant Peripheral Nerve Sheath Tumor (MPNST) - 5 cases (56%), embryonal rhabdomyosarcoma (ERMS) - 2 cases (22%), Wilms tumour (WT) - 1 case (11%), undifferentiated sarcoma - 1 case (11%). The median age at tumour manifestation was 9,5 years old (3 - 16 y.o.). Five patients with MPNST died because of tumour progression; other patients are alive without relapse. One patient had two tumours during life - MPNST and undifferentiated sarcoma and died because of MPNST progression inspite of three chemotherapy lines, radiation and target therapy.

Conclusion: Neurofibromatosis type 1 is a cancer predisposition syndrome, so patients diagnosed with NF1, need special observation in order not to miss tumour development. In case a malignant tumour occurs, patients have to take special treatment according to diagnosis. The most aggressive tumour and one of the most common reasons of death in patients with NF1 is MPNST, that supposes the necessity of searching for new treatment options for these patients.