CNS Embryonal Tumor with BRD4-LEUTX Fusion

CNS embryonal tumors are a group of rare and aggressive brain tumors that primarily affect children. The term “embryonal” refers to the fact that these tumors arise from cells that are normally present during fetal development.

BRD4-LEUTX fusion is a genetic alteration that has been identified in a subset of CNS embryonal tumors. This fusion occurs when the BRD4 gene on chromosome 19 and the LEUTX gene on chromosome 2 are fused together, resulting in a new fusion gene.

The exact role of the BRD4-LEUTX fusion in the development and progression of CNS embryonal tumors is not yet fully understood. However, it is thought that fusion may play a role in promoting the growth and proliferation of tumor cells, as well as in preventing these cells from undergoing programmed cell death (apoptosis).

Patients with CNS embryonal tumors with BRD4-LEUTX fusion may require a different treatment approach than those with other types of CNS embryonal tumors. The optimal treatment plan will depend on a variety of factors, including the patient's age, overall health, and the specific characteristics of the tumor. Treatment options may include surgery, radiation therapy, chemotherapy, and targeted therapies.

Lebrun et al. reported a pediatric case of a novel tumor type among the other CNS embryonal tumors classified within the methylation class “CNS Embryonal Tumor with BRD4-LEUTX Fusion”. The patient was a 4-year girl with no previous history of the disease. For a few weeks, she suffered from headaches, vomiting, and mild fever associated with increasing asthenia and loss of weight leading to a global deterioration of health. MRI brain examination revealed a large, grossly well-circumscribed tumoral mass lesion located in the left parietal lobe, contralateral hydrocephalus, and midline shift. Microscopic examination showed a highly cellular tumor with a polymorphic aspect. The majority of the tumor harbored neuroectodermal features composed of small cells with scant cytoplasm and hyperchromatic nuclei associated with small “medulloblastoma-like” cells characterized by syncytial arrangement and focally a streaming pattern. Tumor cells were diffusely positive for Synaptophysin, CD56, INI1, and SMARCA4 associated with negativity for GFAP, OLIG2, EMA, BCOR, LIN28A, and MIC-2. Additional IHC features included p53 protein expression in more than 10% of the tumor's cells and very interestingly, loss of H3K27me3 expression. The Heidelberg DNA-methylation classifier classified this case as “CNS Embryonal Tumor with BRD4:LEUTX Fusion”. RNA-sequencing analyses confirmed the BRD4 (exon 13)-LEUTX (exon 2) fusion with no other molecular alterations found by DNA sequencing. The case report confirmed that a new subgroup of CNS embryonal tumor with high aggressive potential, loss of H3K27me3 protein expression, BRDA4-LEUTX fusion, named “Embryonal CNS tumor with BRD4-LEUTX fusion”, has to be considered in the new CNS WHO classification [1)].

1)
Lebrun L, Allard-Demoustiez S, Gilis N, Van Campenhout C, Rodesch M, Roman C, Calò P, Lolli V, David P, Fricx C, De Witte O, Escande F, Maurage CA, Salmon I. Clinicopathological and molecular characterization of a case classified by DNA-methylation profiling as “CNS embryonal tumor with BRD4-LEUTX fusion”. Acta Neuropathol Commun. 2023 Mar 18;11(1):46. doi: 10.1186/s40478-023-01549-2. PMID: 36934287.

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