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Diffuse Pediatric-type High-grade Glioma Arising in an Ovarian Mature Cystic Teratoma

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Abstract

Immature neuroectodermal tissue can be found in the ovary as part of an immature teratoma or as part of a teratoma with malignant neuroectodermal transformation. Such lesions may closely resemble central nervous system tumors, but their biologic similarity is unclear. We describe an 18-yr-old female who presented with abdominal pain caused by an ovarian mass with widespread metastases. Histology showed a primitive, high-grade tumor arising in the background of a mature teratoma. The tumor was SOX10 positive, with focal expression of GFAP, S100, NSE, and synaptophysin. Molecular analysis demonstrated co-amplification of PDGFRA and KIT, alterations common in high-grade gliomas. By whole-genome methylation profiling, it clustered into the "diffuse pediatric-type high-grade glioma, RTK1 subtype, subclass c" group. Despite progressing through 2 lines of chemotherapy with widespread metastatic disease, she achieved an excellent response to chemotherapy directed toward aggressive germ cell tumors. This case emphasizes the importance of immunohistochemical, genomic, and epigenetic analyses to accurately classify these exceedingly rare tumors and determine the optimal therapy.