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A novel case of peripheral T-cell lymphoma, not otherwise specified, with rhabdoid morphology and INI1 loss

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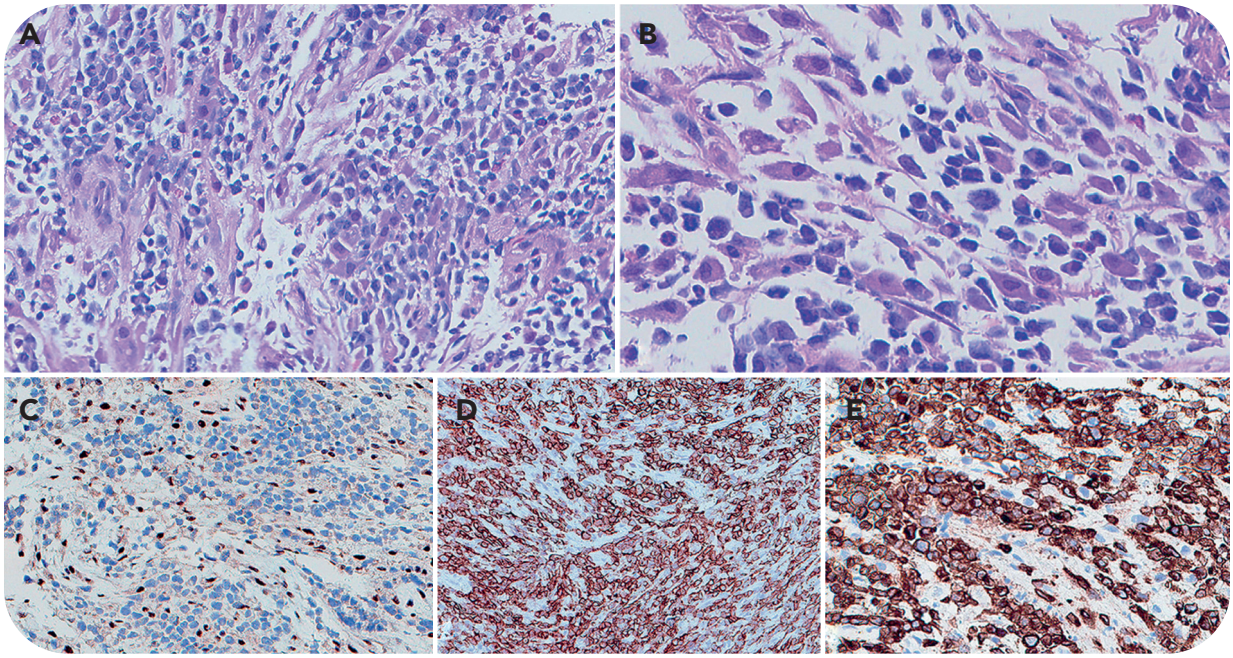

A 6-year-old boy with 22q11.2 distal microdeletion syndrome (characterized by telomeric microdeletions in chromosome 22 encompassing *INI1/SMARCB1* gene and associated with a high incidence of malignant rhabdoid tumors) presented with back pain and vertebral lesions. Vertebral biopsy (panel A; original magnification ×400; hematoxylin and eosin [H&E] stain) showed small to medium cells with compact chromatin with characteristic rhabdoid morphology (abundant eosinophilic cytoplasm and eccentric nuclei) in a subset (panel B; original magnification ×600; H&E stain) and loss of INI1 expression (panel C; original magnification ×400; INI1 immunohistochemistry [IHC] stain). The neoplastic cells were positive for CD5 (panel D; original magnification ×400; CD5 IHC stain), CD3 (panel E; original magnification ×400; CD3 IHC stain), CD2, CD4 (partial weak), CD45, and CD43, and they were negative for CD7, CD8, CD30, anaplastic lymphoma kinase, CD56, CD57, Epstein-Barr encoding region in situ hybridization, and keratin. Ki-67 showed a high proliferation rate. T-cell receptor-γ polymerase chain reaction showed a clonal rearrangement pattern. At 13-month follow-up, the patient was in complete remission after cyclophosphamide, doxorubicin, vincristine, and prednisone therapy, followed by allogeneic hematopoietic stem cell transplant.

This novel peripheral T-cell lymphoma, not otherwise specified, exhibited rhabdoid histomorphology with loss of INI1 expression. Malignant rhabdoid tumors (characterized by rhabdoid cells, loss of INI1 expression, and biallelic inactivation of the *INI1* gene) are described in the central nervous system, kidney, and soft tissue. Limited data exist regarding hematological neoplasms associated with biallelic loss of the *INI1* gene. This case raises awareness of this rare entity and the utility of INI1 immunohistochemistry to facilitate its diagnosis.

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