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Cancer Prone Disease Section

Mini Review

Rhabdoid predisposition syndrome

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Clinics

Note

The following observations have suggested that a new cancer-prone disease, related to the gene hSNF5/INI, could be delineated:
Two siblings with a paravertebral malignant rhabdoid tumor in the first year of life and a poor outcome; no family history;
Renal rhabdoid tumors associated with tumors of the central nervous system in a given patient;
Germ-line mutations of INI1 identified in four children, three with renal rhabdoid tumors and one with an atypical teratoid tumor of the brain (out of 18 atypical teratoid and rhabdoid tumors studied);
And 4 recent pedigrees with - malignant rhabdoid tumor, choroid plexus carcinoma, atypical teratoid tumor, medulloblastoma, and/or primitive neuroectodermal tumor, - either occurring in sibs or in a given patient, - with a INI1 point mutation in the tumor DNA and loss of wild type allele and/or heterozygosity for the mutation in constitutional DNA.

Phenotype and clinics

No apparent stigmata.

Neoplastic risk

Malignant rhabdoid tumors and atypical teratoid tumors, choroid plexus carcinomas, medulloblastomas, and primitive neuroectodermal tumors; highly aggressive tumors; very early onset in children or infants, and, apparently, high penetrance.

Genes involved and proteins

hSNF5/INI1

Location

22q11.2

Mutations

Germinal

Found in this syndrome.

Somatic

Mutation and allele loss events in sporadic rhabdoid tumors, primitive neurectodermal tumors, medulloblastoma, or choroid plexus carcinoma are in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

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