Cancer Prone Disease Section
Short Communication

Rhabdoid tumor
Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: March 1999
Online updated version : http://AtlasGeneticsOncology.org/Tumors/rhabdoidID5037.html

DOI: 10.4267/2042/37521

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.

© 1999 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Classification
- Primarily described as rhabdoid tumor of the kidney (RTK), further extended to tumors of other primary sites: extrarenal rhabdoid tumor (ERRT, or malignant extrarenal rhabdoid tumor MERT)
- They present a wide histological, ultrastructural, and immunocytochemical spectrum.
- May represent a heterogeneous group of neoplasms and also invite confusion with other renal or extrarenal neoplasms, of which is the Favorable Histology Wilms' tumor (with a fair prognosis).
- Finally "composite" extrarenal rhabdoid tumors (CERT) with a recognizable "parent" neoplasm admixed with MERT appear to be of various origin.
- The recent finding that hSNF5/INI1 is involved in true rhabdoid tumors is of paramount importance in this context.

Clinics and pathology

Embryonic origin
Uncertain histiogenesis.

Epidemiology
RTK occurs in infancy and early childhood, median age is 11 mths; unbalanced sex ratio (1.5M/1F); ERRT have been observed in a broader range of patient ages.

Clinics
Often located in the kidney, may occur in various anatomic sites, such as the central nervous system or soft tissues.

Prognosis
Highly aggressive; 80% mortality rate with frequent metastases, predominantly pulmonary; a large study 10 yrs ago found a better outcome for girls (> 50% survival) than for boys (10%).

Cytogenetics

Cytogenetics Morphological
- Normal karyotype or 22q11.2 involvement in a t(Var; 22)(-;q11.2) or in del(22q).
- Loss of heterozygosity (LOH) on chromosome 22; LOH can also occur at chromosome band 11p15.5, indicating that a second gene may also be involved in addition in a subset of rhabdoid tumors.

Genes involved and proteins

hSNF5/INI1
Location
22q11.2

Germinal mutations
Found in the rhabdoid tumor predisposition syndrome.

Somatic mutations
Mutation and allele loss events in sporadic rhabdoid tumors are in accordance with the two-hit model for neoplasia, as is found in retinoblastoma.

References


This article should be referenced as such: