Malignant rhabdoid tumor

A malignant rhabdoid tumor (MRT) is a very aggressive form of tumor originally described as a variant of Wilms' tumour, which is primarily a kidney tumor that occurs mainly in children in 1978.

Malignant rhabdoid tumors (MRT) are a rare and highly malignant childhood neoplasm. Later rhabdoid tumors outside the kidney were reported in many tissues including the liver, soft tissue, and the central nervous system. Several cases of primary intracranial MRT have been reported since its recognition as a separate entity in 1978. The term “rhabdoid” was used due to its similarity with rhabdomyosarcoma under the light microscope. The exact pathogenesis of MRT is unknown.

The cerebellum is the most common location for primary intracerebral MRT (i.e., AT/RT). Biggs et al. were the first to report a primary intracranial MRT around 1987.

Although the cell of origin is not known, cytogenetic studies have suggested an everyday genetic basis for rhabdoid tumors regardless of location with abnormalities in chromosome 22 commonly occurring.

The vast majority of rhabdoid tumors present with a loss of function in the SMARCB1 gene, also known as INI1, BAF47 and hSNF5,, a core member of the SWI/SNF chromatin-remodeling complex. Recently, mutations in a second locus of the SWI/SNF complex, the SMARCA4 gene, also known as BRG1 were found in rhabdoid tumors with retention of SMARCB1 expression. Familial cases may occur in a condition known as rhabdoid tumor predisposition syndrome (RTPS). In RTPS, germline inactivation of one allele of a gene occurs. When the mutation occurs in the SMARCB1 gene, the syndrome is called RTPS1, and when the mutation occurs in the SMARCA4, gene it is called RTPS2. Children presenting with RTPS tend to develop tumors at a younger age, but the impact that germline mutation has on survival remains unclear. Adults who carry the mutation tend to develop multiple schwannomas. The diagnosis of RTPS should be considered in patients with RT, especially if they have multiple primary tumors and/or in individuals with a family history of RT. Because germline mutations result in an increased risk of carriers developing RT, genetic counseling for families with this condition is recommended [1)].

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Sredni ST, Tomita T. Rhabdoid Tumor Predisposition Syndrome. Pediatr Dev Pathol. 2014 Dec 10. [Epub ahead of print] PubMed PMID: 25494491.