The diagnosis of these tumours is confirmed by loss of expression of 1 of 2 components of the SWI/SNF chromatin-remodeling complex, SMARCB1 (also known as INI1) and SMARCA4 (also known as BRG1), due to inactivating mutations in either of these genes. Tumours that retain the expression of INI-1 but that exhibit characteristic morphological features should be tested for loss of BRG-1 expression by immunohistochemistry.

The diagnosis of 'rhabdoid tumour predisposition syndrome' should be considered in presence of multiple primary tumours, and/or in individuals with a family history of rhabdoid tumour. Genetic counselling for families with this condition is recommended.

Multimodality therapy combining surgical excision, chemotherapy and radiotherapy is the mainstay of treatment. Extrarenal rhabdoid tumours are highly aggressive with extensive metastasis and poor survival rates.[4] Few studies suggest a better prognosis in older children, regardless of primary tumour location.[5]

When a young patient presents with an aggressive soft tissue tumour which shows a rhabdoid morphology and polyphenotypic immunoprofile, an extrarenal rhabdoid tumour should be considered in the differential diagnosis and Immunohistochemical expression of INI-1 should be evaluated.

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