

SMARCB1 in Atypical teratoid rhabdoid tumor

Identification of loss of [SMARCB1](#)/INI1 expression in poorly differentiated (PD) [chordoma](#) in pediatric patients suggests that PD chordoma is an entity molecularly distinct from conventional chordoma or [Atypical teratoid rhabdoid tumor](#), which is also characterized by loss of SMARCB1/INI1 expression by inactivating mutation of the SMARCB1/INI1 gene. So far, around 20 cases of pediatric PD chordoma with loss of SMARCB1/INI1 expression have been reported.

Cha et al. report two cases of pediatric PD chordoma with loss of SMARCB1/INI1 expression, which is very rare among the pediatric chordoma types. Both patients presented clival masses on preoperative MRI. Histologically, both tumors had nonclassic histologic features for conventional chordoma: sheets of large epithelioid to spindle cells with vesicular nuclei and prominent nucleoli. Both cases revealed nuclear expression of brachyury, loss of SMARCB1/INI1 expression and lack of embryonal, neuroectodermal, or epithelial component. One case showed heterozygous loss of [EWSR1](#) gene by break-apart fluorescence in situ hybridization that reflected loss of SMARCB1/INI1 gene. Based on the clival location and histologic findings along with the loss of SMARCB1/INI1 expression and positivity for nuclear brachyury staining, the final pathologic diagnosis for both cases was PD chordoma ¹⁾.

In a study, Torchia et al analyzed 191 primary [Atypical teratoid rhabdoid tumor](#) ATRTs and 10 ATRT [cell lines](#) to define the genomic and epigenomic landscape of ATRTs and identify subgroup-specific therapeutic targets. They found ATRTs segregated into three epigenetic subgroups with distinct genomic profiles, [SMARCB1](#) genotypes, and chromatin landscape that correlated with differential cellular responses to a panel of signaling and epigenetic inhibitors. Significantly, they discovered that differential methylation of a PDGFRB-associated enhancer confers specific sensitivity of group 2 ATRT cells to [dasatinib](#) and [nilotinib](#), and suggest that these are promising therapies for this highly lethal ATRT subtype ²⁾.

References

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