About a third of soft tissue tumors are recognized by gene fusions. Some of these fusions are not histotype specific and occur in entities with totally different clinicopathological features. A good example is the fusion between EWSR1 and genes of the CREB transcription factor gene family (CREB1 or ATF1) [1].

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 [2].

1)
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2)
Cha YJ, Hong CK, Kim DS, Lee SK, Park HJ, Kim SH. Poorly differentiated chordoma with loss of SMARCB1/INI1 expression in pediatric patients: A report of two cases and review of the literature. Neuropathology. 2017 Aug 15. doi: 10.1111/neup.12407. [Epub ahead of print] PubMed PMID: 28812319.