Coffin-Siris syndrome (CSS) is a rare congenital disorder with variable clinical phenotype consisting of developmental delay and characteristic facial features. It is caused by mutations in the chromatin remodeling switch/sucrose nonfermenting complex. Although SWI/SNF genes are widely implicated in tumorigenesis, only 8 cases of neoplasm have been reported in patients with CSS.

A case of anaplastic astrocytoma (WHO grade III) in an 18-month-old child with Coffin-Siris syndrome (CSS) due to a de novo germline missense SMARCE1 mutation. Additional molecular features of the tumor are described as well. The role of missense SMARCE1 mutations in tumor predisposition in children with CSS should be further investigated to better inform genetic counselling [1)].

1)
Lin B, Kesserwan C, Quinn EA, Einhaus SL, Wright KD, Azzato EM, Orr BA, Upadhyaya SA. Anaplastic Astrocytoma in a Child With Coffin-Siris Syndrome and a Germline SMARCE1 Mutation: A Case Report. J Pediatr Hematol Oncol. 2018 Nov 28. doi: 10.1097/MPH.0000000000001361. [Epub ahead of print] PubMed PMID: 30499906.