Desmoid tumor

Desmoid tumors, also known as aggressive fibromatosis, are locally infiltrating musculoaponeurotic neoplasms arising in connective tissues. Desmoid tumors may be associated with familial adenomatous polyposis (FAP), a genetic disorder that presents with hundreds to thousands of precancerous colorectal polyps.

Li et al., report the case of an 18-month-old boy who underwent resection of a right temporal desmoid tumor (initially diagnosed as cranial fasciitis) and developed a bilateral frontoparietal calvarial desmoid tumor 2 years later. The patient underwent gross-total resection of the tumor that required a large cranioplasty. He was subsequently diagnosed with FAP. The patient has been without tumor recurrence for 9 years afterwards and has not required revision of his cranioplasty. This is the first report describing a recurrent cranial desmoid tumor in a pediatric patient with FAP. The authors believe, however, that some of the cases previously reported as cranial fasciitis are likely desmoid tumors pathobiologically and genetically [1)].

A 2-year-old girl presented with a rapidly enlarging tumor of the forehead and a family history of FAP. The tumor was cultured for cytogenetic studies. A DNA linkage study using flanking and intragenic polymorphisms of the adenomatous polyposis coli (APC) gene was performed to identify the allele loss in the tumor. Germline mutation identification was by single strand conformation polymorphism analysis of exon 15 of the APC gene, with subsequent double stranded sequencing of fragments with conformational changes. A mutation-induced loss of a restriction site was used to confirm allele loss in the tumor.

Microscopically, the tumor had desmoid features. Cytogenetic analysis of the tumor demonstrated loss of chromosome region 5(q21q22). A truncating adenomatous polyposis coli (APC) gene mutation was identified in the leukocyte DNA from the child and her affected father. Linked DNA markers suggested that the tumor had lost the maternal, wild-type allele. A mutation-induced restriction endonuclease site alteration demonstrated hemizygosity of the mutant sequence in the tumor DNA [2)].

1)
Li L, Jensen JN, Szabo S, VanTuinen P, Lew SM. Recurrent giant cranial desmoid tumor in a 3-year-old boy with familial adenomatous polyposis requiring bifrontoparietal cranioplasty: case report. J Neurosurg Pediatr. 2016 Dec;25(6):703-707. PubMed PMID: 27635978.
2)
de Silva DC, Wright MF, Stevenson DA, Clark C, Gray ES, Holmes JD, Dean JC, Haites NE, Dunlop MG. Cranial desmoid tumor associated with homozygous inactivation of the adenomatous polyposis coli gene in a 2-year-old girl with familial adenomatous polyposis. Cancer. 1996 Mar 1;77(5):972-6. PubMed PMID: 8608492.