Optic nerve pilomyxoid astrocytoma

Pilomyxoid astrocytoma (PMA) is a described tumor that typically occurs in the chiasmatic-hypothalamic region in young children and has unique histopathologic and clinical characteristics. These tumors have been previously diagnosed as pilocytic astrocytoma (PA). PMA appears to have a higher rate of recurrence and CSF dissemination than typical PA.

Pilomyxoid astrocytoma might be an infantile form of pilocytic astrocytoma and speculate that a subset of pilomyxoid astrocytomas in the optic pathway/hypothalamus originates from the optic chiasm, possibly derived from radial glia existing in the embryonic optic chiasm.

Snuderl et al. found that although subtypes of optic gliomas are indistinguishable on imaging, the microvascular network of pilomyxoid astrocytoma, a subtype of optic glioma with abundant myxoid matrix, is characterized by the presence of endothelium-free channels in the myxoid matrix. These tumors show normal perfusion by clinical imaging and lack histological evidence of hemorrhage organization or thrombosis. The myxoid matrix is composed predominantly of the proteoglycan versican and its linking protein, a vertebrate hyaluronan and proteoglycan link protein 1. They propose that pediatric optic gliomas can maintain blood supply without endothelial cells by using invertebrate-like channels, which they termed primitive myxoid vascularization. Enzymatic targeting of the proteoglycan versican/hyaluronan and proteoglycan link protein 1 rich myxoid matrix, which is in direct contact with circulating blood, can provide novel therapeutic avenues for optic gliomas of childhood [1].

Noonan syndrome (NS; MIM 163950) is an autosomal dominant syndrome which is clinically diagnosed by the distinct facial features, short stature, cardiac anomalies and developmental delay. About 50% of cases are associated with gain of function mutations in PTPN11 gene which leads to activation of the RAS/mitogen-activated protein kinase signaling pathway. This is known to have a role in tumorigenesis. Despite this, only limited reports of solid tumors (Fryssira H, Leventopoulos G, Psoni S, et al. Tumor development in three patients with Noonan syndrome. Eur J Pediatr 2008;167:1025-1031; Schuettpelz LG, McDonald S, Whitesell K et al. Pilocytic astrocytoma in a child with Noonan syndrome. Pediatr Blood Cancer 2009;53:1147-1149; Sherman CB, Ali-Nazir A, Gonzales-Gomez I, et al. Primary mixed glioneuronal tumor of the central nervous system in a patient with Noonan syndrome. J Pediatr Hematol Oncol 2009;31:61-64; Sanford RA, Bowman R, Tomita T, et al. A 16 year old male with Noonan's syndrome develops progressive scoliosis and deteriorating gait. Pediatr Neurosurg 1999;30:47-52) and no prior reports of optic gliomas have been described in patients with NS. We present here a patient with NS with a PTPN11 mutation and an optic pathway pilomyxoid astrocytoma [2].

Edwards et al. presented an unusual radiographic appearance of a pilomyxoid astrocytoma in an 11-year-old child. Preoperative images suggested a dural-based, homogenously enhancing lesion coupled with an enlarged optic nerve. Surgery revealed an intraparenchymal lesion of the right temporal lobe. There was hyperintensity on T2 MRI sequences, suggesting infiltration of the tumor along the optic tracts [3].

Terasaki et al. reported on a 5-year-old boy with an approximately 2-month history of progressively worsening loss of vision. Radiographic studies with contrast revealed an enhanced mass within the optic nerve, an enhanced lesion in the leptomeninges, and diffusely scattered nonenhanced white matter lesions in the craniospinal axis. The patient was treated with a 10-week carboplatin and vincristine regimen without a biopsy. After completing induction and 1 maintenance cycle, however, the patient developed coma caused by hydrocephalus. External ventricular drainage was performed and a biopsy was taken through ventriculoscopy, revealing PMA. The patient was then treated with craniospinal irradiation and concomitant temozolomide, a regimen to which he had a complete response. Two years after initial presentation the patient was free of disease [4].

References

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