Rare intracranial EWSR1-rearranged myxoid mesenchymal tumour in a teenager

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DESCRIPTION
A 14-year-old girl presented with 10 months of worsening headaches with associated nausea and 1 month of visual impairment, including blurry and double vision. Her headaches initially occurred monthly but increased in frequency and became intractable to medical management. Physical examination was significant for papilloedema on funduscopic examination and a right inferior quadrantanopia but was negative for other neurological abnormalities. MRI revealed a peripherally located, homogeneously hyperenhancing mass in the left posterior supratentorial compartment that was T1-hypointense and T2-hyperintense with slight heterogeneity (figure 1). Cortical vasculature displacement indicated extra-axial location. No dural tail or calcifications were appreciated, but hyperintensity of the white matter involving the left parietal lobe and compression of the left occipital lobe. The neuroradiographic differential diagnoses included meningioma, atypical haemangioepithelioma, lymphoma and gliosarcoma. The patient underwent surgical resection where neuropathology revealed spindle cell proliferation of low cellularity with angulate, hyperchromatic nuclei and thin strands of cytoplasm separated by an abundant myxoid background (figure 2). Sparsely scattered rosette-like structures with a central core of densely eosinophilic, hypocellular collagenous material were detected. Little mitotic activity was observed, and reticulin staining revealed abundant network deposition around the tumour cells. The histological differential diagnoses for myxoid soft tissue include myxoma, myxoid liposarcoma, myxoid chondrosarcoma, low-grade fibromyxoid sarcoma and myxoid undifferentiated pleomorphic sarcoma (myxofibrosarcoma). Immunohistochemistry showed diffuse and strongly positive staining for CD99 and slight epithelial membrane antigen and desmin positivity. Few (<1%) Ki-67+ nuclei were noted, and a subset of tumour nuclei stained positive for beta-catenin. Scattered CD68+ cells throughout the lesion were noted. Immunostaining was negative for S100, SMA, synaptophysin, GFAP, CD34, STAT-6, NF, myogenin and ALK-1. Features customarily observed in angiomatoid fibrous hystiocytosis (AFH)—fibrous pseudocapsule, lymphoplasmacytic infiltrates and blood-filled cystic spaces were not appreciated. Next-generation sequencing detected a variant of unknown significance in TOP2A (c97A>C) but no known clinically significant variants. Microarray likewise detected no clinically significant abnormalities. Fluorescence in situ hybridisation demonstrated rearrangement of the EWSRI gene at 22q12. These findings were most consistent with a diagnosis of EWSR1-rearranged myxoid mesenchymal tumour. The patient underwent complete tumour resection and has been...
myxoid mesenchymal tumour.

Genetic rearrangement featuring $EWSR1$ fusion can occur in a diverse array of mesenchymal neoplasms. However, $EWSR1$-rearrangement mesenchymal tumours in the central nervous system are exceedingly rare and were unreported until recently, with the presentation of a small case series of adolescents and young adults with intracranial myxoid mesenchymal tumours positive for $EWSR1$ rearrangement. Subsequent literature has sought to determine whether these tumours are a novel neoplastic entity or represent a myxoid variant of AFH, but has not been able to definitively establish their status. The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

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REFERENCES


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