Rare presentation of juvenile xanthogranuloma in the posterior fossa of a toddler

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DESCRIPTION

A 13-month-old girl presented to the emergency department with 1 month of progressively enlarging right neck mass not responsive to oral antibiotics. In addition, she had decreased appetite, fussiness, poor eye contact, unsteady crawling and night sweats. Prior to presentation, her birth history was unremarkable and she had no other medical problems. She had normal development and a normal neurological examination though the family noted that her gross motor function was more unsteady in the month leading up to her presentation.

MRI of the head and neck revealed a large, heterogeneous extra-axial posterior fossa mass with associated restricted diffusion and cerebellar compression with mass effect on the fourth ventricle and brainstem (figure 1). Differential considerations included a primitive neuroectodermal tumour, atypical teratoid/rhabdoid tumour, haemangiopericytoma, paraganglioma or glioblastoma. However, there was erosion of the calvarium and extracranial extension of the mass along the right inferior occipital calvarium and enhancing right cervical lymph nodes, which raised the suspicion for a non-primary central nervous system (CNS) neoplasm such as Langerhans cell histiocytosis (LCH). MRI of the entire spine assessing for leptomeningeal involvement was normal. CT of the chest, abdomen and pelvis showed multiple, bilateral conspicuous axillary lymph nodes and a right upper paratracheal node without other evidence of visceral metastatic disease. The patient underwent complete tumour resection with biopsies of the posterior fossa mass as well as the surrounding skull. Permanent sections showed spindled ovoid cells with vesicular chromatin and distinct nucleoli. Among the spindled cells are large number of multinucleated giant cells with scattered Touton forms, and smaller numbers of foamy histiocytes, eosinophils, lymphocytes and plasma cells, consistent with juvenile xanthogranuloma (JXG).[1] Immunohistochemistry showed strongly positive CD68+giant cells (figure 2) and additional testing was performed to rule out LCH. Tumour cells were 10%–20% Ki67 positive, INI-1 negative, CD163 positive, factor XIIIa positive, anaplastic lymphoma kinase 1 negative and Langerin negative. Tumour cytogenetics was positive for 46,XX,ins(8;12)(q24.3;q24.3q15),del(12)(q13)(20).

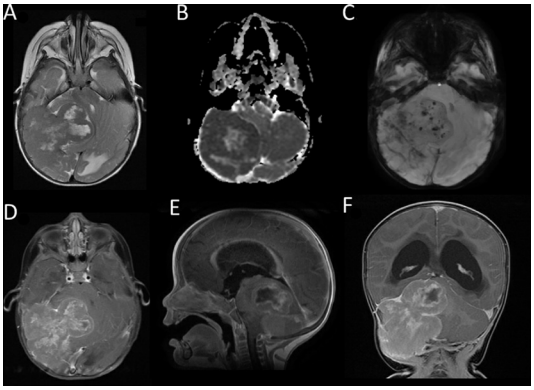

Figure 1 MRI brain demonstrating a large heterogeneous mass in the right posterior fossa (A) with associated diffusion restriction (B) and microhaemorrhage on susceptibility weighted imaging. (C) Postcontrast studies show heterogeneous enhancement throughout the mass (D) with mass effect on the fourth ventricle, pons and medulla with slightly downward placement of the cerebellar tonsils (E, F).

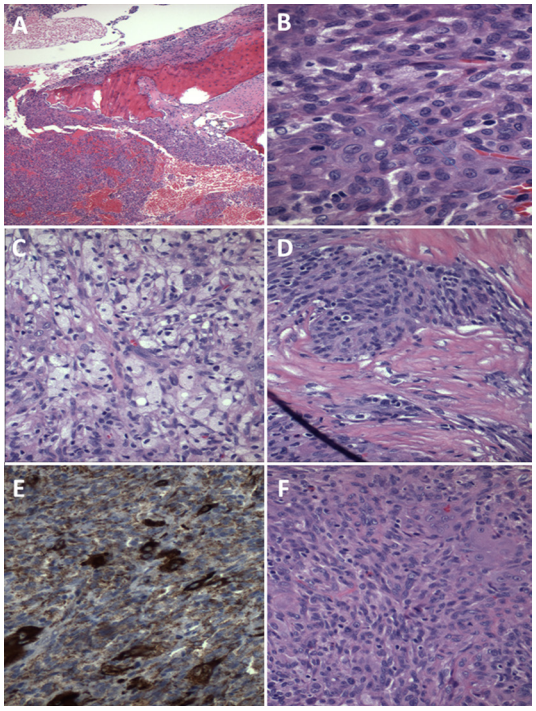

Figure 2 Light microscopy and immunohistochemistry showing (A) H&E 40×: histiocytes and spindle cell tumour infiltrating bone; (B) H&E 400×: histiocytic-appearing tumour cells with oval nuclei, small nucleoli and moderate amount of cytoplasm and a mitotic figure; (C) H&E 400×: foamy histiocytes present focally; (D) H&E 200×: nest of histiocytic cell surrounded by dense collagen; (E) CD68 staining 200×: giant cells strongly positive for CD68 with weak staining of background cells; and (F) H&E 200×: histiocytic cells with small numbers of eosinophils.

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JXG is a rare, non-LCH of childhood, with 75% of cases presenting before 1 year of age. The lesions are most commonly cutaneous papules or nodules of the scalp, neck and trunk that self-resolve over the course of several years. Ocular disease is rarer, and involvement of the CNS and disseminated disease is unusual.[1] JXG has also been reported in patients with neurofibromatosis type 1 (NF1) and juvenile chronic myelogenous leukaemia.[2] Interestingly, the patient's maternal great-grandfather had NF1, though there were no other family members with cutaneous lesions and the patient's NF1 gene testing was negative. Although JXG in the CNS is rare, lesions in the suprasellar region, cerebellopontine angle and spinal cord have been reported.[3–5] Thus, the clinical spectrum of JXG, especially extracutaneous involvement, is broad and should be considered on the differential for paediatric posterior fossa tumours. Further studies with expanded population studies and cytogenetics are needed to determine which subtypes are susceptible to a more aggressive clinical course.

Learning points

- Juvenile xanthogranuloma (JXG) is a non-Langerhans cell histiocytosis seen mostly in childhood that rarely involves the central nervous system.
- JXG should be on the differential for a young child presenting with a posterior fossa tumour with or without disseminated disease.

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