INTRACRANIAL JUVENILE XANTHOGRANULOMA IN A CHILD: A CASE REPORT

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Background. Juvenile xanthogranuloma (JXG) is a rare non-Langerhans cell histiocytic disorder that predominantly affects children. Cutaneous involvement is the most common presentation, whereas central nervous system (CNS) involvement is uncommon and poses significant diagnostic challenges. Intracranial JXG may mimic primary brain tumors and other pathological conditions; therefore, accurate diagnosis requires integration of clinical, neuroimaging, histopathological, and immunohistochemical findings.

Objective. To present a rare case of intracranial juvenile xanthogranuloma in a child and to describe its clinical presentation, diagnostic features, treatment, and clinical course.

Materials and Methods. We report the case of a 7-year-old girl with progressive neurological symptoms lasting approximately five months. Clinical findings, brain magnetic resonance imaging (MRI), surgical treatment, histopathological and immunohistochemical findings, as well as cytogenetic and molecular investigations were analyzed.

Results. The patient presented with headache, progressive weakness of the left upper and lower extremities, inability to walk independently, nausea, vomiting, dysphagia, and generalized weakness. Brain MRI demonstrated a right hemispheric intra-axial mass with extensive surrounding vasogenic edema and associated mass effect. The patient underwent craniotomy with surgical resection of the intracranial lesion.

Histopathological examination demonstrated histiocytic proliferation composed of macrophages and multinucleated giant cells. Immunohistochemical analysis revealed diffuse expression of CD68 and CD163, focal expression of Factor XIIIa and S100, and negative staining for CD1a. These morphological and immunophenotypic findings were consistent with juvenile xanthogranuloma. Molecular testing by fluorescence in situ hybridization (FISH) demonstrated a KIAA1549::BRAF rearrangement in 48% of tumor cells, whereas ALK rearrangement was not confirmed.

The patient subsequently received systemic therapy with cytarabine and cladribine in combination with the MEK inhibitor cobimetinib. A transient clinical improvement was observed during treatment; however, the disease subsequently progressed, with rapid deterioration of the patient's general and neurological condition and ultimately a fatal outcome.

Practical Recommendations. Juvenile xanthogranuloma should be considered in the differential diagnosis of intracranial mass lesions in pediatric patients, particularly in cases with atypical neurological manifestations and absence of characteristic cutaneous lesions. Accurate diagnosis requires correlation of neuroimaging with histopathological and immunohistochemical findings. In patients with atypical or aggressive disease, molecular characterization may provide additional information regarding potentially relevant alterations involving the MAPK signaling pathway.

Keywords: *juvenile xanthogranuloma; non-Langerhans cell histiocytosis; central nervous system; intracranial lesion; pediatric neuro-oncology; KIAA1549::BRAF.*