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PLEOMORPHIC XANTHOASTROCYTOMA- A RARE ENTITY

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Abstract

INTRODUCTION: Pleomorphic xanthoastrocytoma (PXA) is an exceedingly rare astrocytic tumor, accounting for less than 1% of these neoplasms and under 0.3% of primary CNS tumors. It primarily affects children and adolescents, most commonly in the temporal lobe, where it often presents as epilepsy. PXAs are slow-growing and typically lack significant peritumoral edema. Histologically, PXAs are marked by pleomorphic tumor cells including spindle cells, polygonal cells, multinucleated giant cells and lipid-laden cells, with nuclear inclusions and eosinophilic granular bodies.

CASE REPORT: A 9-year-old female presented with a one-month history of persistent headaches and vomiting, and two episodes of convulsions. MRI revealed a 4.2 x 3.9 x 6 cm heterogeneously enhancing intra-axial mass in the right fronto-temporo-parietal region, with mild to moderate vasogenic edema and a contralateral midline shift. The tumor was resected via right fronto-parietal craniotomy and histopathology confirmed pleomorphic xanthoastrocytoma. Immunohistochemistry showed IDH-1 negativity, while FNAC suggested high-grade glioma. Gene mutation showed BRAF (V600E) positivity.

DISCUSSION: Pleomorphic xanthoastrocytoma is a rare, well-circumscribed astrocytoma that commonly presents as seizures. Although typically cerebral, PXAs have also been reported in less common locations like the cerebellum, spinal cord and retina.

CONCLUSION: PXA has a favourable prognosis, with a five-year survival rate of approximately 70%, though outcomes vary based on tumor grade, molecular profile, age and treatment response.

KEYWORDS: Pleomorphic xanthoastrocytoma, BRAF, Astrocytic tumors

INTRODUCTION

Pleomorphic xanthoastrocytoma (PXA) is an exceedingly rare tumor, constituting less than 1% of all astrocytic neoplasms and accounting for under 0.3% of primary central nervous system tumors[1]. The term PXA was given by Kepes in 1979. Later in 1993, it was included under 'circumscribed astrocytic tumors' in the WHO classification system of tumors of central nervous tumors as a WHO grade 2 tumor[1]. Predominantly, it affects the younger individual, particularly children and adolescents(1st-2nd decade of life) with no gender predilection[2]. PXA may occur in any of the four lobes of the brain, with the predilection of temporal lobe where they present as temporal lobe epilepsy[2]. These tumors exhibit slow growth generally without significant peritumoral oedema.

Approximately, 98% of PXA are localised to cerebrum, with nearly half of these arising in the temporal lobe. Histopathologically, the tumor cells are highly pleomorphic comprising spindle cells, polygonal cells, multinucleated giant cells, lipid-laden cells alongside the frequent presence of eosinophilic granular bodies[3]. The marked variability in nuclear size and presence of nuclear inclusions further contribute to characteristic pleomorphism. PXA exhibit BRAF mutation.

CASE PRESENTATION

Here we present a case of a 9-year old female who presented with a one-month history of persistent headache and vomiting accompanied by two episodes of convulsions over the preceding week. MRI (P+C) of brain revealed a welldemarcated, lobulated, heterogeneously enhancing intra-axial mass, approximately measuring 4.2 x 3.9 x 6 cm, located in the right fronto-temporo-parietal region (Figure.1,2). The lesion was associated with mild to moderate vasogenic edema and exerted mass effect, resulting in a contralateral midline shift to the left. A right fronto-parietal craniotomy was performed and the excised tumor measuring 5 x 4 x 2 cm was submitted for histopathological evaluation. Grossly, the tumor appeared multiple fragments of greyishwhite tissue (Figure.3). Microscopically features consistent with pleomorphic xanthoastrocytoma were identified (Figure.4a,4b,4c,4d).

Immunohistochemistry revealed IDH-1 negativity (Figure.5) . Gene mutation showed BRAF (V600E) positivity. Squash cytology suggested the diagnosis of a high grade glioma.

DISCUSSION

Pleomorphic xanthoastrocytoma is a highly unusual type of astrocytoma. These lesions are generally well-circumscribed astrocytoma and may present as solid or cystic masses. Clinically, they most commonly present as seizures[4]. Although predominantly found in cerebral cortex, there have been rarereports of PXA in cerebellum, spinal cord, meninges and retina. Some PXAs show increased mitotic activity (more than equal to 5 mitosis/10hpf with or without areas of necrosis called as 'PXA with anaplastic features'[5]. Although, PXAs are considered as low grade gliomas, it can be associated with malignant transformation, recurrence and death[6]. PXA has also been diagnosed in paediatric patients with a histological diagnosis of PXA were identified[5,7]. Majority of the tumors were cystic components and associated with peritumoral edema[5,7]. About 2/3rds of PXA harbour BRAF mutation[8]. A study showed BRAF V600E positive in 12 of 20% (60%) WHO grade 2 PXA, a percentage more than anaplastic PXA and glioblastoma arising in a PXA[8].

CONCLUSION

Despite its rarity, pleomorphic xanthoastrocytoma is associated with a favourable prognosis. Studies have reported a five-year survival rate of approximately 70%. Prognosis however is contingent upon several factors including tumor grade, molecular profile, age and treatment response[9]. Surgery is the mainstay of treatment. PXA has the potential to reoccur, hence radiotherapy is given for recurrence. BRAF inhibitors are given in BRAF mutation positive patients[9].

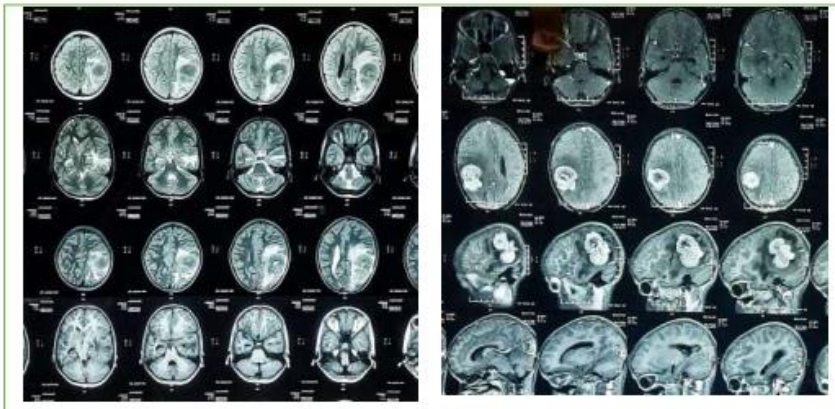

Figure.1,2 : C-MRI (BRAIN) – Lesion shows T1 heterogenous hypointense, T2/Flair heterogenous hyperintense

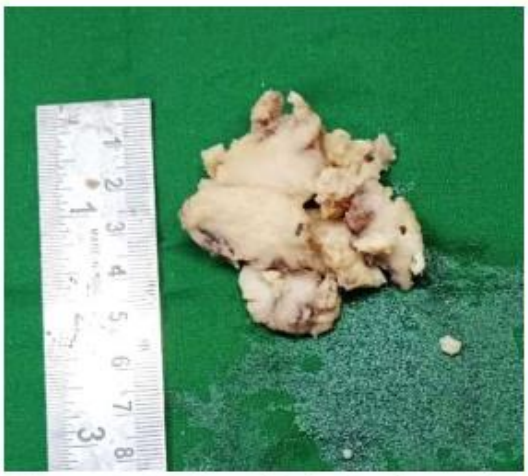

Figure. 3: GROSS-Multiple bits of greyish-white tissue altogether measuring 5.5x4.7x2cm.

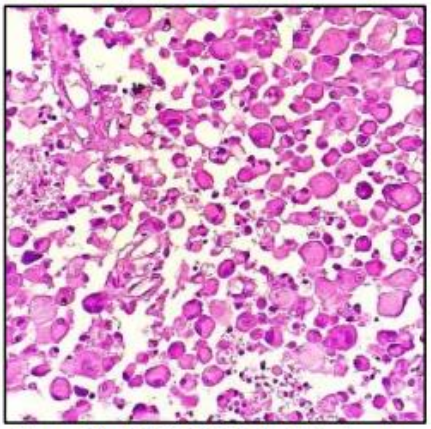

Figure.4(a)- H&E (400x): PXA showing abundant foamy lipidised cells.

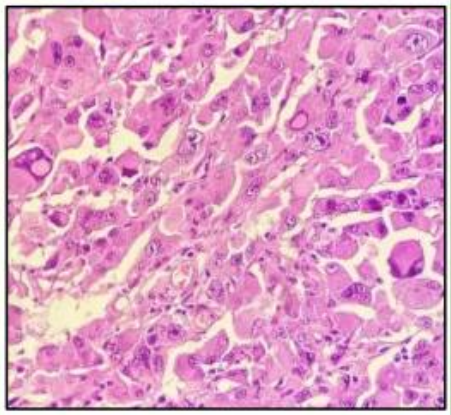

Figure.4(b)- H&E (400x): PXA showing numerous nuclear inclusions.

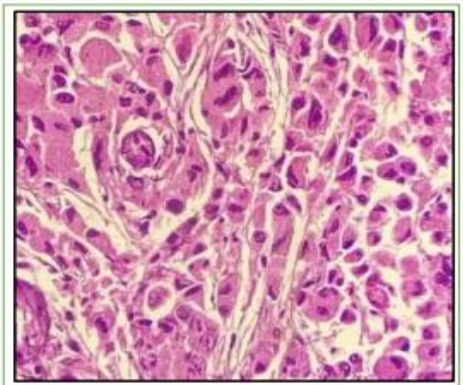

Figure.4(c)- H&E (400x):PXA showing spindle cells and eosinophilic granular bodies.

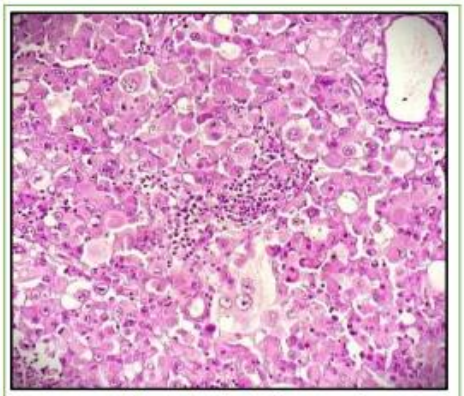

Figure.4(d)- H&E (400x):PXA showing focal perivascular lymphocytic infiltration.

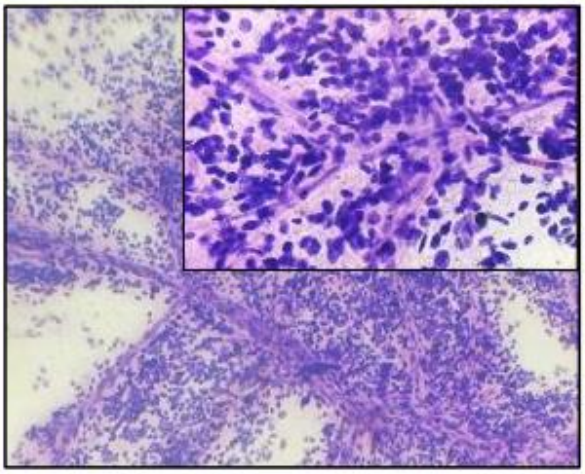

Figure.5: H&E *40x* (with inset,400x): Squash showing pleomorphic tumor cells present over a glial fibrillary background

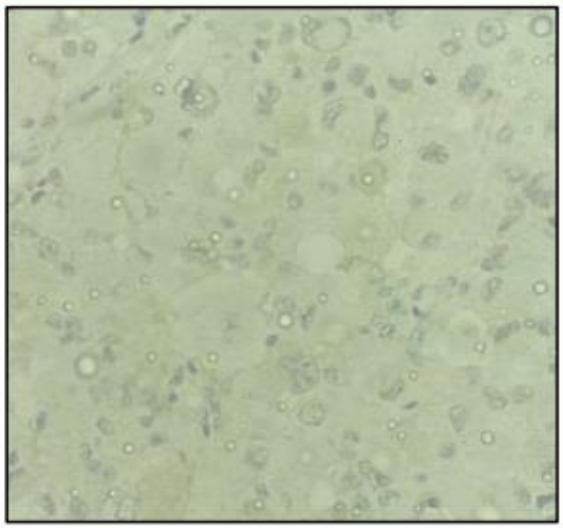

Figure. 6: IHC showing immunonegative for IDH-1.

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