A stitch in time saves sight: Craniopharyngioma

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ABSTRACT

Craniopharyngiomas are rare dysontogenic, extra-axial, slow-growing tumors arising from the remnants of the Rathke's pouch and located within the sellar or parasellar region, which can be solid, cystic, or calcified. We are reporting a case of an 11-year-old female child who presented with complaints of sudden onset rapid diminution of vision in the right eye more than the left eye for 2 months, for distant and near vision, with normal visual fields. The patient was diagnosed as having craniopharyngioma based on magnetic resonance imaging. Surgery was done without delay, restoring her vision. Histopathology of tumor confirmed the diagnosis. This case highlights the fact that immediate neuroimaging in children presenting with sudden onset, rapidly progressive vision loss helps in early diagnosis and management of craniopharyngioma and to retrieve lost vision.

Key words: *Craniopharyngioma, Magnetic resonance imaging, Surgical management, Visual loss*

Craniopharyngiomas are rare, relatively benign neoplasms that typically arise in the sellar/suprasellar region. They are derived from embryonic cellular remnants of the Rathke's pouch found along the path of the primitive adenohypophyseal and craniopharyngeal duct. The prevalence of craniopharyngioma is 0.13–2 per 100,000 [1]. It occurs due to a defect in Wnt signaling pathway reactivation due to B catenin gene mutation. Craniopharyngiomas are primarily suprasellar tumors (75%) while a small intrasellar component is present in 20–25% of cases.

Occasionally, craniopharyngiomas appear as intraventricular, homogeneous, soft-tissue masses without calcification (papillary subtype). The third ventricle is a particularly common location. Rare or ectopic locations reported include nasopharynx, posterior fossa, and extension down the cervical spine. The tumor has a bimodal distribution. The first peak occurs between the ages of 5 and 15 years and a second smaller peak occurs in adults aged over 40 years [1]. Clinical presentation is varied on account of the variable location and size of the tumor. Presenting complaints include headache, nausea, vomiting, visual symptoms, and endocrine dysfunction.

CASE REPORT

An 11-year-old female child born of non-consanguineous marriage with normal birth history and developmental milestones, presented with complaints of sudden onset, progressive rapid diminution of vision in the right eye more than the left eye for 2 months and intermittent headache. Initially, the child complained of blurred vision for distant vision. There was no history of specific visual field loss. Two months later, she could not identify coins or see numbers on the mobile. Family history was not significant.

On general examination, the patient's vitals were normal. On neurological examination, higher functions were normal. Examination of the second cranial nerve showed visual acuity of finger counting at 2 feet and 3 feet in the right and left eye, respectively. However, visual fields were normal on the ophthalmological examination. Other cranial nerves were normal. There was no motor or sensory deficit or cerebellar signs.

Multiplanar magnetic resonance imaging (MRI) brain showed thin-walled predominantly cystic sellar-suprasellar lesion, measuring 3 × 3.6 × 4 cm and pituitary gland was not seen. These features were suggestive of craniopharyngioma (Fig. 1). Hormonal assays showed low cortisol level - one microgram/dl (normal range = 1–22 microgram/dl) and thyroid-stimulating hormone (TSH) 5.8 Mu/L (normal range = 0.55–5.31 Mu/L).

The patient was treated immediately. Surgical management was done with total excision of the tumor and the excised specimen sent for histopathology. Histopathology of the tumor revealed solid and cystic areas with cells arranged in large islands and sheets with multiple foci of calcifications suggestive of adamantinomatous craniopharyngioma WHO Grade 1. The patient was also started on steroids 1 mg/kg/day in view of low cortisol and oral thyroxine 25 microgram once daily in the morning (empty stomach) in view of hypothyroidism. Postoperatively, vision recovered completely. Post-operative MRI showed complete resection of tumor.

DISCUSSION

Craniopharyngioma is a relatively rare benign slow-growing tumor which has a prevalence of 0.13–2 per 100,000 [1]. The first description of a craniopharyngioma was given in 1857 by Zenker.

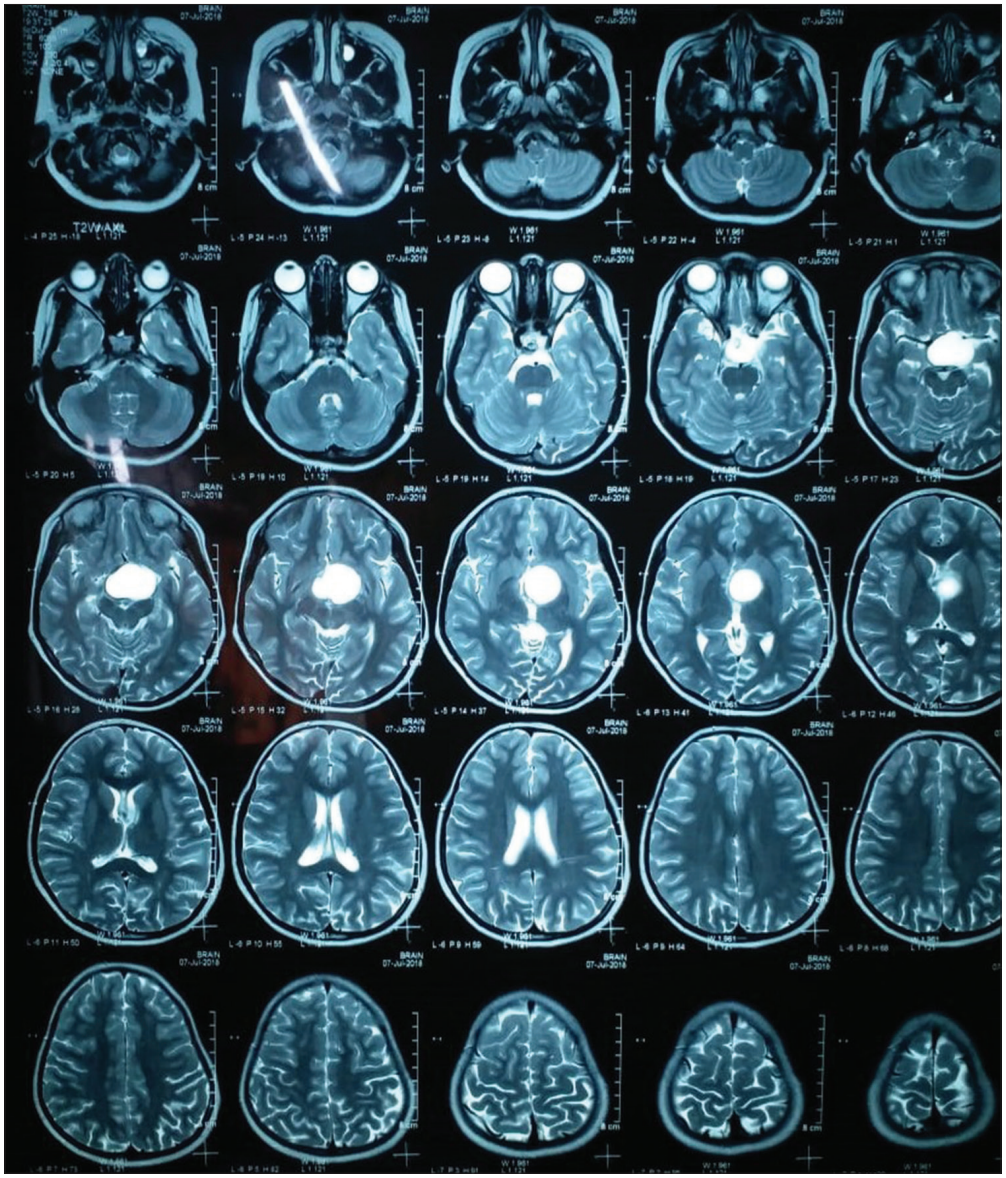

Figure 1: Magnetic resonance imaging brain (plain) showing craniopharyngioma

The term craniopharyngioma was introduced by Cushing in 1932. Thereafter, a detailed study was done and its histological subtypes were identified. The diagnosis of this tumor is based on clinical presentation and radiological findings.

Craniopharyngioma presents with visual disturbances, increased intracranial pressure, neurobehavioral and endocrine problems. In a study by Wan *et al*., [2] the most common presenting features were headache (76%), nausea or vomiting (32%), and vision loss (31%). Our patient presented with visual disturbance and intermittent headache without any nausea or vomiting. Various reports have described neurological disturbances such as headache, vision loss, and visual field defects, along with manifestations of endocrine deficiency such as growth retardation and delayed puberty to be the most common presenting symptoms of craniopharyngiomas [3-6]. Although our case presented with a diminution of vision, there was no visual field defect. These tumors can stretch the diaphragma sellae and cause headaches [7]. Obstruction of the cerebral aqueduct and the foramen of Monro may also lead to hydrocephalus, which makes a shunt necessary [7,8]. Hydrocephalus was not seen in our patient.

At diagnosis, endocrine dysfunction is found in up to 80% of patients [9]. Reduced growth hormone secretion is the most frequent endocrinopathy and can be present in up to 75% of patients [3,4]. This is followed by follicle-stimulating hormone/luteinizing hormone deficiency, which can be seen in 40% of patients and then adrenocorticotropic hormone and TSH deficiency in 25% [3]. Our patient had low cortisol levels and hypothyroidism which is a less commonly seen endocrinopathy in craniopharyngioma. A study by DeVile *et al*. states that hypoadrenalism and associated hypoglycemia contribute to morbidity and mortality in children with craniopharyngioma with endocrine dysfunction [10].

The diagnosis on MRI is mainly based on the three characteristic components of the tumor: Cystic, solid, and calcified. The cystic component constitutes the most important part of the tumor and shows variable signal depending on the chemical-physical

properties of its content [11]. Fluid content will appear hypointense in T1-weighted and hyperintense in T2-weighted images, whereas lipid (due to cholesterol), methemoglobin, or protein content will appear as hyperintense in T1 and T2 sequences. The solid portion has an isointense signal in T1- and a hyperintense signal in T2-weighted images with enhancement after gadolinium, at variance with the cystic component. However, contrast enhancement is not a consistent feature. Calcifications can appear as areas of low signal in all sequences but are generally visualized better with computerized tomography scans [12,13]. In our patient, T2 hypointensities with blooming on gradient recalled echo images were seen in the sella and of the lesion wall. No calcifications were seen on MRI in our patient.

Differential diagnosis includes pituitary adenoma, eosinophilic granuloma, vascular malformations (aneurysms), and congenital malformations like Rathke's cleft cyst [1,3]. In small intrasellar or enclosed tumors, total resection is done which is most easily achieved, and adjunctive radiotherapy is not required. Radiotherapy is required in cases of incomplete tumor removal, which occurs frequently with extrasellar craniopharyngiomas [7]. Near-total excision of the tumor by sparing the hypothalamus, carotid arteries, and visual apparatus, followed by fractionated radiotherapy, provides the best hope of low long-term morbidity and longer survival [14]. In our case, total excision of the tumor was done and no adjunct radiotherapy was required.

There are two histological subtypes of craniopharyngioma – adamantinomatous and papillary. The adamantinomatous subtype is most commonly seen in children between 5 and 15 years and is commonly associated with Wnt pathway aberrations; however, it can also be present sometimes in adults over 40 years [1]. In our patient, histopathology of tumor showed adamantinomatous subtype. The papillary subtype is found almost exclusively in adults which are associated with BRAF V 600 mutation. However, Borrill *et al*. reported papillary craniopharyngioma in a 4-year-old girl with BRAF V600E mutation [15].

Regardless of the approach, the incidence of endocrine dysfunction is high following surgical treatment. Hyperfractionated multiportal stereotactic radiotherapy and gamma knife radiosurgery are promising therapeutic adjuncts to standard radiotherapy, due to their potential ability to reduce treatment-associated morbidity in this condition [3]. In children, however, the benefit of any additional radiotherapy treatment should be balanced against the relatively high risk of inducing hypopituitarism later in life. Long-term follow-up is advised as 15% of craniopharyngiomas recur. Management involves a multidisciplinary team of pediatricians, pediatric neurologists, neurosurgeons, endocrinologists, and ophthalmologists.

CONCLUSION

Craniopharyngioma remains a challenging tumor because despite its benign histological appearance, it is often associated with unfavorable and occasional disastrous sequelae. Hence, a high index of suspicion in children presenting with visual symptoms can lead to early diagnosis and treatment. Immediate neuroimaging and surgery in our patient with craniopharyngioma, who presented with sudden onset and rapidly progressive vision loss, resulted in complete retrieval of lost vision. Early diagnosis with neuroimaging and immediate surgical treatment is essential for a good outcome. However, endocrine abnormalities require treatment and long-term monitoring even after surgery. All children with craniopharyngioma should be managed and followed up by a multidisciplinary team.

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