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Case Report

Primary anterior visual pathway germinoma in a 13-year-old boy: A case report

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ABSTRACT

Background: Primary optic nerve and chiasmal germinomas are very rare. These lesions can commonly be mistaken for optic pathway gliomas based on imaging alone. It is radiosensitive and cured in most of the cases.

Case Description: We report a rare case of a 13-year-old boy with primary bilateral optic nerves and chiasmal germinoma who underwent partial surgical resection followed by radiotherapy. Follow-up brain imaging after two months post-radiotherapy showed interval regression of the tumor. Our literature review identified that 12 reported cases of primary anterior visual pathway germinoma had been reported to regress significantly post-radiotherapy alone or with chemotherapy.

Conclusion: Histologic correlation is essential for appropriate treatment, alleviating symptoms, and avoiding irreversible vision loss.

Keywords: Chiasmal germinoma, Germ cell tumors, Intracranial germinoma, Optic nerves germinoma

INTRODUCTION

Intracranial germ cell tumors (GCTs) comprise <1% of all central nervous system (CNS) tumors[1] and 3.8% of pediatric brain tumors.[18] germinoma is a more frequent tumor type of intracranial GCTs that occurs in the pediatric age group.[1,8] The pineal, suprasellar, basal ganglia regions and thalamus are the commonly reported locations of germinoma.[11] Primary germinoma of the optic nerve or chiasm is extremely rare, with limited reported cases in the literature.[2-4,6,9,16,20-22] The primary anterior visual pathway germinoma diffusely infiltrates the optic nerve or chiasm, which impacts the prognosis of these cases.[2-4,6,9,16,20-22] Distinguishing primary anterior visual pathway germinoma from glioma based on radiologic features has proved to be challenging; moreover, the treatment regimens differ between primary anterior visual pathway germinoma and glioma, hence why tissue diagnosis is crucial.[13,21] The literature demonstrates that intracranial germinoma has favorable outcomes compared to other pediatric malignant tumors.[1] In this case report, we present a case of a 13-year-old boy diagnosed with primary anterior visual pathway germinoma by discussing associated clinical and radiological findings and reviewing related published literature.

CASE DESCRIPTION

A 13-year-old boy who was born to second-degree consanguineous parents was not known to have any medical illness. He presented to the emergency department of King Faisal Specialist Hospital and Research Center at Jeddah with a persistent headache for four weeks, followed by progressive visual loss for over two weeks associated with polyurea before the presentation. The parents denied having a similar condition or history of any malignancies in the family. On examination, the pupils were dilated bilaterally with a sluggish reaction in the left eye. The ophthalmologic assessment revealed diminished visual acuity with no light perception in the right eye and light perception OS. A fundoscopic examination showed bilateral optic atrophy. No skin stigmata or café-au-lait spots were inspected. A brain magnetic resonance imaging (MRI) revealed a lobulated enhancing lesion arising primarily from the optic chiasm involving the pre-chiasmatic optic nerves and the hypothalamus, measuring 3.1 × 3.7 × 1.7 cm (anteroposterior × mediolateral × craniocaudal dimensions).

The infundibulum was normal. Radiologic findings were most suggestive of optic pathway glioma [Figure 1]. The hormonal assay revealed central hypothyroidism, central hypoadrenalism, central diabetes insipidus (DI), and hyperprolactinemia. The patient was evaluated by a pediatric endocrinologist for hormonal replacement therapy. The patient underwent a right pterional craniotomy and a Transylvanian approach to the optic chiasm and nerves. Intraoperatively, the optic chiasm and bilateral optic nerves diffusely thickened with a fleshy appearance. A tumor biopsy was obtained, followed by lesion debulking for size reduction. Postoperative recovery was uneventful. The patient was discharged home in stable clinical condition. The specimen exhibited histological patterns in keeping with germinoma [Figure 2]. The whole spine MRI showed no drop metastatic lesion. Serum level of alpha-fetoprotein and beta-human chorionic gonadotropin was normal. The case was discussed in a multidisciplinary tumor board with the conclusion of starting SIOP CNS GCT 96 protocol, with four cycles of carboplatin and etoposide alternating with etoposide and ifosfamide, followed by radiation therapy after evaluation. During the first cycle, the patient developed a significant reaction to etoposide in terms of chest tightness, rash, and low blood pressure. As the patient was unfit for chemotherapy, the treatment plan consisted of solely adjuvant radiotherapy. He received a total of 4600 centigray (cGy) in 29 fractions. After completion of the radiotherapy sessions, the brain MRI revealed an interval reduction in the size of optic chiasm thickening without residual enhancing lesion [Figure 3]. On the follow-up visit after two months from radiotherapy, no vision improvement was noted; otherwise, he was well.

DISCUSSION

GCTs classified by the 2021 World Health Organization classification of the tumors of the CNS into mature teratoma, immature teratoma, teratoma with somatic-type malignancy, germinoma, embryonal carcinoma, yolk sac tumor, choriocarcinoma, and mixed GCT.[14] Intracranial GCTs are usually diagnosed at 10–21 years old.[11] Germinoma comprises 65–76% of all intracranial GCTs.[1,11] The majority of intracranial GCTs occur in pineal (48%), suprasellar (37%), and basal ganglia-thalamus (3%) regions; it may occur intracranially within the ventricle.[11] Primary intracranial GCTs have been reported in rare locations such

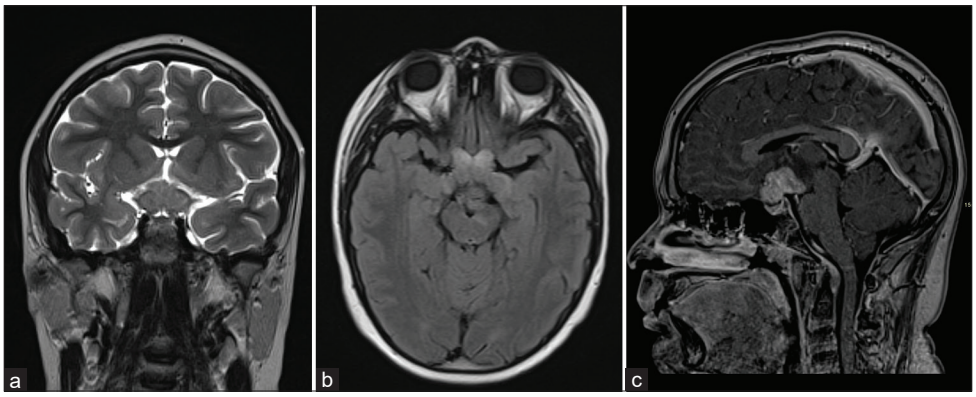

Figure 1: (a) Coronal T2-weighted image shows lobulated thickening of the optic chiasm with normal pituitary infundibulum and (b) axial fluid-attenuated inversion recovery shows the involvement of pre-chiasmatic optic nerves and optic tracts, (c) midsagittal contrast-enhanced T1-weighted spoiled gradient recalled echo demonstrates the avid lesion enhancement with involvement of the hypothalamus.

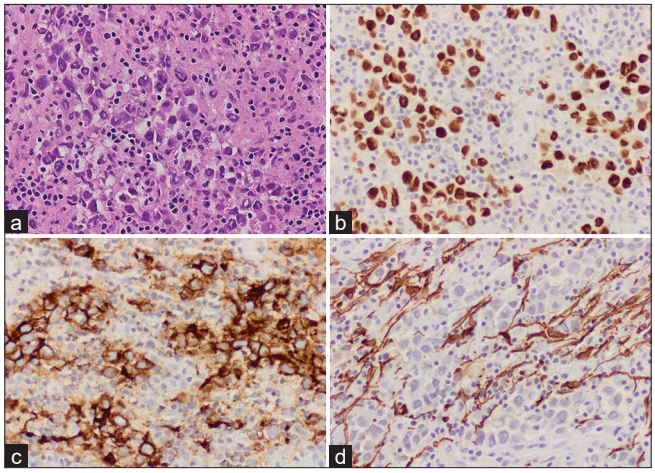

Figure 2: (a) Hematoxylin and eosin stained section (H&E @ ×20), showing nests of slightly discohesive, large epithelioid cells with abundant cytoplasm and pleomorphic nuclei intermixed with benign lymphocytes, (b) immunohistochemistry reveals positive immunoreactivity against SALL4, OCT4, (c) immunohistochemistry reveals positive immunoreactivity against placental alkaline phosphatase (PLAP), and (d) negative immunohistochemistry for glial fibrillary acidic protein (GFAP), S100, AE1/AE3, CD45, and CD30.

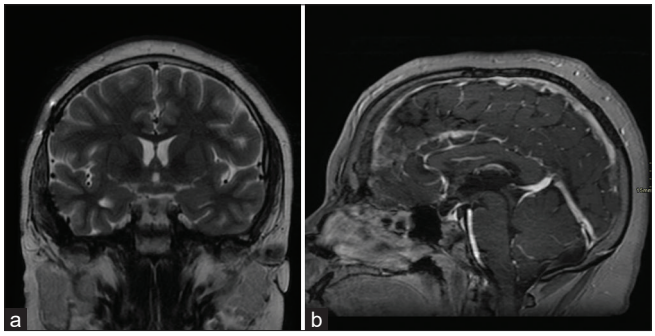

Figure 3: (a) Coronal T2-weighted image shows interval reduction in the size of optic chiasm thickening (b) no residual enhancing lesion on midsagittal contrast-enhanced T1-weighted magnetic resonance image.

as the cerebellum, brainstem, and optic nerve.[4,9,15-17] One of the pathogenesis theories that explain intracranial GCTs is a disruption of primordial germ cell migration control where dislocation is intracranially followed by malignant transformation.[7] The neurological symptoms and signs depend on the lesion's location. Most of the suprasellar germinoma present initially with DI.[2] Furthermore, the triad of visual deterioration, DI, and hypopituitarism are common in suprasellar lesion.[2,13] Bowman and Farris, in 1990, were the first who reported a case of primary chiasmal germinoma in a 44-year-old man based on tumor biopsy, who was cured completely by radiotherapy.[2] According to our literature search, there are 12 reported cases of primary anterior visual pathway germinoma, most of these cases were male, three cases were female [Table 1].[2-4,6,8,9,12,16,20-22] The mean age of diagnosis of all reported cases including our case was 21 years. The painless progressive vision deterioration was the main complaint of all reported cases. About 58% (n = 7) for those with chiasmal germinoma presented with DI. Hormonal abnormalities were reported in 66% of the anterior visual pathway germinoma. The literature demonstrates that only 3.9% of the optic pathway, chiasmal, and hypothalamic gliomas present with DI compared to 60% of the optic pathway and chiasmal germinomas, which largely affects the primary imaging/preoperative differential.[2-5,10,12,16,20-22] Moreover, most of the reported cases presented with the triad of visual deterioration, DI, and hypopituitarism. The lesion, when small and solely chiasmal, can be missed on a non-enhanced computed tomography (CT) scan of the brain; thus, an MRI of the brain becomes essential to assess intracranial involvement.[2] The imaging features of anterior visual pathway germinoma can largely overlap with those of optic pathway gliomas on CT and MRI. Suprasellar germinomas typically arise from the hypothalamus, showing extension into the pituitary infundibulum. Both lesions can show variable signal intensity on the different pulse sequences, heterogeneous enhancement, and lack of calcifications. A study by Panyaping *et al*. showed that the significant difference in apparent diffusion coefficient (ADC) values could be utilized as a distinguishing feature between suprasellar germinomas and chiasmatic/hypothalamic gliomas, germinoma histological features demonstrate high cellularity with densely packed cells whereas low cellularity is observed in chiasmatic/hypothalamic gliomas. Thus, suprasellar germinoma had a lower average ADC value compared to the minimum ADC value in chiasmatic/hypothalamic gliomas.[19] It is important to distinguish between germinoma and other differential diagnoses of anterior visual pathway lesions because the treatment protocol and prognosis are different.[21] Obtaining tissue diagnosis is necessary.[4] All formerly reported cases underwent surgical procedures for tumor biopsy. The prognosis is better in germinoma when compared to other GCTs.[22] Radiotherapy is the cornerstone in the management of localized intracranial germinoma, while chemotherapy can be utilized in disseminated disease.[13,16] The survival rate is 91% for those who are treated with radiotherapy at 5 and 10 years.[5] The primary anterior visual pathway germinoma has been reported to regress significantly post-radiotherapy alone or with chemotherapy in reported cases, including our case.

Table 1: Summary of anterior visual pathway germinoma reported cases.

Author	Age	Sex	Presentation	Examination	Location	MR imaging findings	Postop therapy
Current Case	13 y	M	Vision loss, DI, and hypothyroidism	Bilateral optic nerve atrophy	Chiasm+bilateral optic nerves	Diffuse high-intensity signal of optic chiasm extended to optic nerves	RT
Geurten *et al*.	14 y	F	Vision loss, DI, and hypothyroidism	L optic atrophy, R mild optic disc pallor	Chiasm+L optic nerve	Heterogenous enhanced L optic nerves and chiasm	RT+Chemo
Chaudhry *et al*.	25 y	F	Vision loss and prolactin elevation	Bitemporal hemianopsia	Chiasm	Enhanced bilateral optic nerves and chiasm	RT
Xia *et al*.	30 y	F	Vision loss, DI, secondary amenorrhea, and galactorrhea	NLP in L eye, only R nasal field intact in R eye	Chiasm	Heterogeneous, ring enhancing lesion of optic chiasm	RT
Rath *et al*.	15 y	M	Vision loss	R optic disc pallor, infiltrative lesion in L optic disc	Chiasm+bilateral optic nerves	T1 isointense thickened of optic nerves extend to chiasm	RT
Diluna *et al*.	11 y	M	Vision loss, DI, hypopituitarism, and precocious puberty	Bilateral optic nerve atrophy	Chiasm+bilateral optic nerves	Heterogeneous, ring enhancing lesion of optic chiasm and optic nerves	RT+Chemo
	22 y	M	Vision loss, DI, and hypothyroidism	R homonymous visual field loss	Chiasm+L optic nerve	Heterogenous enhanced L optic nerve and chiasm	RT+Chemo
Nadkarni *et al*.	11 y	M	L vision loss	L optic atrophy	L optic nerve	Enhanced and thickened of L optic nerve	RT
Krolak-Salmon *et al*.	35 y	M	Vision loss	Bilateral optic disc pallor	Chiasm+R optic nerve	Moderate enhanced R optic nerve and chiasm	-
Iizuka *et al*.	31 y	M	Vision loss	-	R optic nerve	-	-
Wilson *et al*.	9 y	M	Vision loss, DI and hypothyroidism	Bilateral pale optic discs; R temporal VF defect; L centronasal VF	Chiasm+bilateral optic nerves	high-intensity signal of optic chiasm and optic nerves	RT
Bowman and Farris	44 y	M	Vision loss, DI, hypotestosteronism and hypothyroidism	Bilateral optic nerve atrophy	Chiasm	Diffuse high-intensity signal of optic chiasm	RT

DI: Diabetes insipidus, y: Years, L: Left, R: Right, M: Male, F: Female, RT: Radiotherapy, Chemo: Chemotherapy, NLP: No light perception, VF: Visual field, RT: Radiotherapy

CONCLUSION

Primary intracranial optic and chiasmal germinoma should be considered in the differential diagnosis of optic pathway lesions. Brain imaging alone can be suboptimal in differentiating between anterior visual pathway germinoma and gliomas. Histologic correlation is essential for appropriate treatment, alleviating symptoms, and avoiding irreversible vision loss.

Ethical approval

King Faisal Specialist Hospital and Research Center Institutional review board approved the case report. Study Number: IRB 2020-CR-25.

Declaration of patient consent

Patient's consent not required as patient's identity is not disclosed or compromised.

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Nil.

Conflicts of interest

There are no conflicts of interest

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The author(s) confirms that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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