Bilateral optic neuropathy with central diabetes insipidus in a child

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Central nervous system germ cell tumors are rare and they occur in the first two decades of life.[1] Optic nerve germinomas can sometimes mimic optic nerve inflammation.[2] In this case report, we discuss an 11-year-old girl who presented with features of presumed bilateral optic neuritis and developed polyuria and polydipsia, subsequently she was diagnosed to have infiltrative etiology. Her clinical and radiological presentations were initially consistent with inflammatory optic neuropathy. Poor visual recovery to steroid therapy and progressive visual loss warranted the need for optic nerve biopsy which revealed germinoma.

Key words: Demyelinating diseases, diabetes insipidus, germinoma, optic neuritis

Optic nerve germinomas can be misdiagnosed as optic neuritis. Herein, we report a child suspected to have bilateral optic nerve inflammation but subsequently diagnosed as infiltrative optic neuropathy.

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_281_18

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Manuscript received: 26.02.18; Revision accepted: 06.08.18

Case Report

An eleven-year-old child presented with complaints of sudden onset painless vision loss in both eyes over 6-week duration. She was suspected to habe bilateral retrobulbar optic neuritis and treated with intravenous methylprednisolone therapy for 3 days followed by oral steroids taper 3 weeks back elsewhere. She had undergone complete serological work up-hemogram, NMOIgG Ab, Quantiferon TB, VDRL,

Cite this article as: Selvakumar A, Muthumeena M, Noronha OV, Bapu S. Bilateral optic neuropathy with central diabetes insipidus in a child. Indian J Ophthalmol 2018;66:1642-4.

RA, ANA, ANCA, ACE, lysozyme, and Mantoux skin test, which were all negative prior. On examination, her best corrected visual acuity was 1/60, less than N 36 in right eye, and 6/18, N6 in left eye. Both pupils reacted sluggishly to light with relative afferent pupillary defect in the right eye. Color vision was reduced in both eyes. Fundus examination revealed pale disc in both eyes. Humphrey visual field with size 5 target of both eyes revealed advanced field loss right more than left eye. Visual evoked potentials of both eyes showed a delayed latency and reduced amplitude more in the right than the left eye. Magnetic resonance imaging (MRI) brain and orbit with gadolinium contrast revealed thickened right optic nerve and bulky chiasm with postcontrast enhancement [Fig. 1]. While the patient was on tapering oral steroids, she had developed skin lesions due to varicella zoster infection. Cerebrospinal fluid examination did not show any abnormal cells. She was treated with IV immunoglobulin and acyclovir. On her 2-month follow-up, her visual acuity further dropped to perception of light in right eye and 1/60 in left eye. Apart from a history of polyuria and polydipsia, her referral neurological examination was normal. Repeat lumbar puncture revealed increased CSF protein. By then her vision had dropped to no perception of light in both eyes. She was suspected to have progressive demyelinating disorder – NMOSD – ON (Neuromyelitis optica spectrum disorder associated optic neuritis) and received a course of intravenous methylpredninsolone therapy and a dose of intravenous rituximab 500 mg. However, her serum NMO IgG antibody was negative, her brain and spine MRI did not reveal any demyelinating lesions. But, repeat MRI brain and orbits revealed increase in size and thickening of bilateral optic nerves and chiasm with contrast enhancement when compared with previous imaging [Fig. 2]. As she had progressive optic neuropathy with no response to therapy and increase in optic nerve and chiasm thickening, optic nerve biopsy was done. Biopsy revealed a poorly differentiated malignant tumor, probably germinoma [Fig. 3]. She was advised for radiotherapy and her polyuria and polydipsia improved following oral desmopressin.

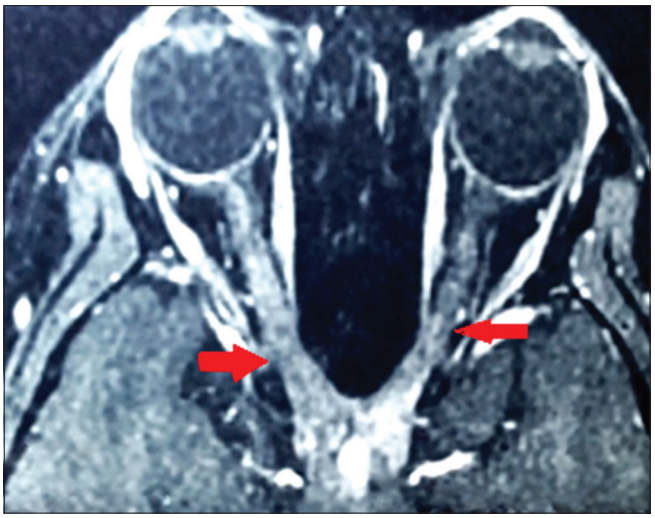

Figure 1: Magnetic resonance imaging brain and orbits axial cut showing T2 hyperintense signal in the bilateral optic nerves with enlarged intracranial optic nerves and chiasm with infundibulum thickened

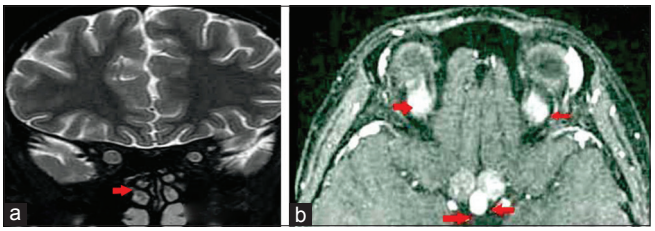

Figure 2: (a) Repeat magnetic resonance imaging – coronal cut showing bulky chiasm and thickened infundibulum; (b) magnetic resonance imaging brain and orbits with gadolinium contrast axial cut showing moderate increase in severity of bilateral optic nerves and optic chiasm signal intensities and thickening with prominent and nodular anterior lobe of pituitary gland

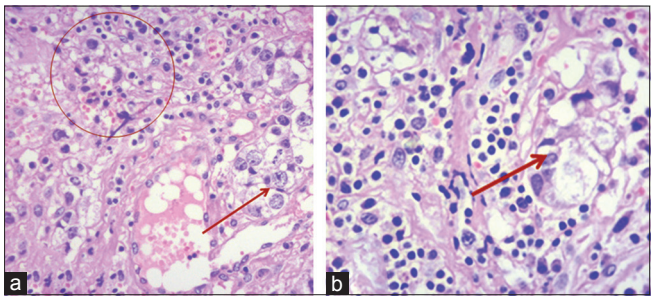

Figure 3: Histopathological examination of optic nerve biopsy: (a) image showing mitotic figures (round mark) and tumor cells (arrow mark) – few cells which were large, rounded with lobules, and had abundant cytoplasm with large, vesicular nuclei, and a prominent nucleoli; (b) image showing tumor cells (arrow mark) in detail along with abundant lympho plasmocytic infiltrates scattered throughout the lesion

Discussion

Intracranial germ cell tumors affect children and young adults, with peak incidence of 10–19 years of age. Clinical manifestation depends on the location and size of the tumor and associated endocrine abnormalities.[2] Visual pathway germinomas involving chiasm and optic nerve are rarely reported.[3-7] Clinical and radiological features of intracranial neoplasm can mimic granulomatous inflammation making early diagnosis difficult.[5] Strong inflammatory reaction can accompany germinomas.[7] Suprasellar germinomas often present with diabetes insipidus followed by visual impairment.[8] Infundibular thickening is usually observed as a feature of neurohypophysis germinomas. Due to anatomic proximity of neurohypophysis and optic chiasm, progressive visual decline can occur due to compression, tumor cell invasion, and the associated inflammation. Poor visual recovery following steroid therapy with clinical features of presumed optic neuritis and thickened optic nerve and chiasm on neuroimaging can be seen in granulomatous diseases and NMO-SD associated optic neuropathies. Germinomas are radiologically well delineated, oval or lobular or partly infiltrative, and expansive tumors.[9] Differential diagnosis of bulky chiasm includes inflammation (infundibuloneurohypophysitis, Wegener's, sarcoidosis), neoplasm's (langerhans cell histiocytosis, metastases, brain tumors, leukemia's), or infections (tuberculosis).[10]

Krolak-Salmon *et al*. reported a patient with similar presentation who was treated as having optic neuritis with high-dose corticosteroids.[9]

This case report emphasizes that infiltrative optic neuropathy in a child can be misdiagnosed as demyelinating disorder, but repeated neuroimaging and biopsy are required to clinch the diagnosis. In this case, germinoma masqueraded as inflammatory optic neuropathy.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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