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Case report

Unilateral paraneoplastic optic disc edema and retinal periphlebitis in pineal germinoma

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ABSTRACT

Purpose: To describe a unilateral ocular paraneoplastic syndrome in pineal germinoma.

Observations: A 24-year-old male presented with diplopia, excessive thirst, and frequent urination. Cranial MRI showed a mass in pineal gland. Dorsal midbrain syndrome signs were present. Examination showed optic disc edema and segmental retinal periphlebitis in right and normal fundus in left eye. Rheumatologic work-up was negative. Brain biopsy confirmed pineal germinoma. Retinal findings were attributed to paraneoplastic syndrome. Resolution of optic disc edema and retinal periphlebitis occurred following chemotherapy and focal irradiation.

Conclusions: and importance: To date there are two published case reports on bilateral optic disc edema and retinal periphlebitis occurring as a paraneoplastic syndrome in pineal germinoma. This is the first report on a patient with pineal germinoma who had unilateral paraneoplastic involvement characterized by optic disc edema and retinal periphlebitis and who showed complete resolution of ocular disease after treatment of underlying tumor.

1. Introduction

Central nervous system germ cell tumors are rare heterogeneous group of tumors mostly diagnosed in children and adolescents. Their incidence varies between geographical regions. They account for 2–3% of primary intracranial neoplasms and for 8–15% of pediatric cases from Japan and other Asian countries and for 0.3–0.6% of primary intracranial neoplasms and 3–4% of pediatric cases from Europe and North America. Pineal region is the most frequent affected location followed by the suprasellar region.[1]

Clinical presentation of pineal germinoma is commonly by signs and symptoms of increased intracranial pressure (papilledema, headache, nausea, and vomiting) and visual disturbances. Suprasellar germinoma usually presents with endocrine abnormalities including disruption of hypothalamohypophyseal axis. Most common ocular manifestations of pineal germinomas include signs and symptoms of papilledema and dorsal midbrain syndrome which is characterized by upward gaze palsy, diplopia, and nystagmus due to tectal plate compression.[1,2] While rare, there are two reported cases of a novel paraneoplastic syndrome of patients who presented with bilateral optic disc edema and retinal periphlebitis as a result of pineal germinoma.[3,4] We herein report a case with pineal germinoma who had unilateral ocular involvement characterized by optic disc edema and retinal periphlebitis as paraneoplastic syndrome.

2. Case report

A 24-year-old male patient was referred to the Ophthalmology Department with a preliminary diagnosis of pineal tumor according to cranial MRI findings conducted in July 2016. The patient was complaining of doubling of vision for one and a half week and excessive thirst and frequent urination for three months. Review of systems was negative for autoimmune and autoinflammatory diseases. His medical and family history was unremarkable.

Ophthalmologic examination showed a best corrected visual acuity of 1.0 in both eyes. He had limited upward gaze, convergence nystagmus with attempted gaze, and pupillary light-near dissociation indicating dorsal midbrain syndrome. Biomicroscopy was unremarkable in both eyes. Intraocular pressures were 16 mmHg in both eyes. Fundus examination showed optic disc edema and segmental retinal periphlebitis with no vitreous cell in the right eye (Fig. 1 A). The fundus of the left eye was normal (Fig. 1B). Fluorescein angiography of the right eye

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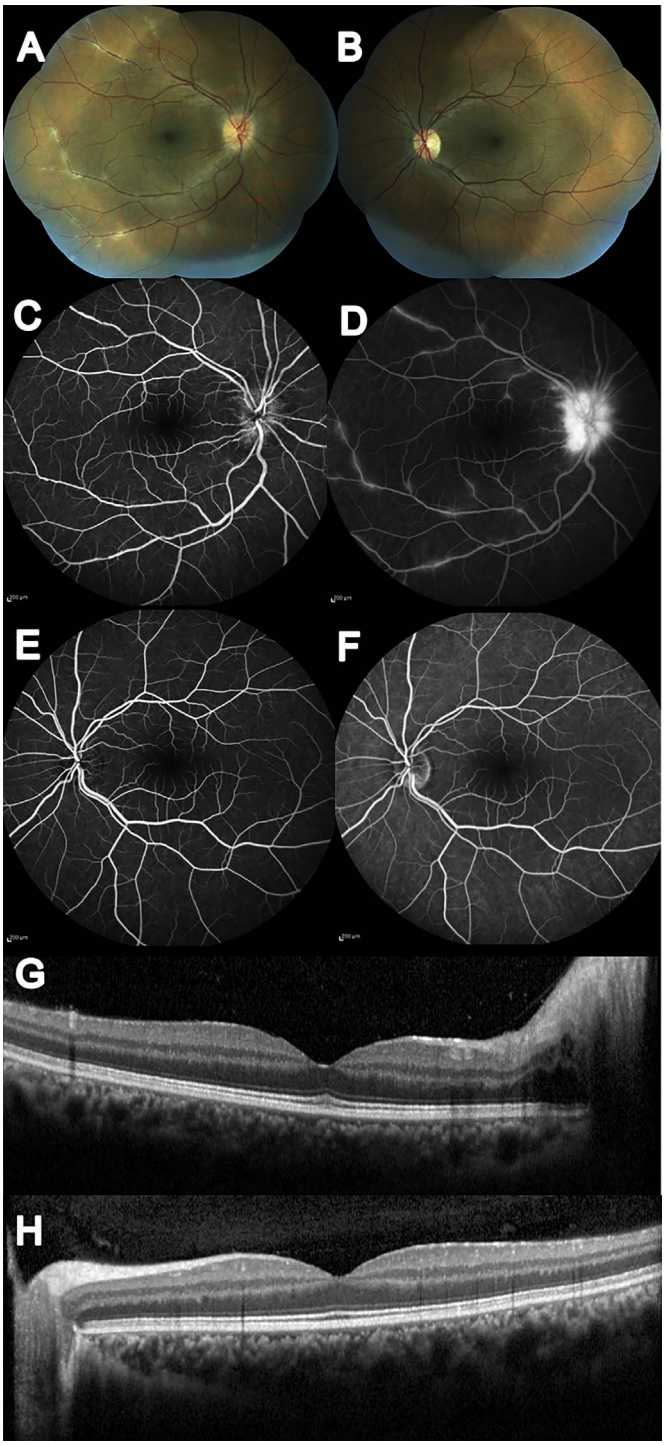

Fig. 1. Color fundus photograph shows optic disc edema and segmental retinal periphlebitis in the right eye (A) and a normal appearance in the left eye (B). Fluorescein angiography of right eye shows mild optic disc staining and faint segmental hyperfluorescence along retinal veins in early phase (C) and staining and leakage from optic disc and retinal veins in late phase angiogram (D). Early (E) and late (F) phase fluorescein angiography is normal in left eye. Spectral-domain optical coherence tomography shows a normal foveal microstructure in the right (G) and left eye (H). (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

revealed mild optic disc staining and faint segmental hyperfluorescence along retinal veins in early phase and staining and leakage from optic disc and from retinal veins in late phase angiogram (Fig. 1C and D). The left eye showed no abnormal angiographic finding in early and late frames of the angiogram (Fig. 1E and F). Spectral-domain optical coherence tomography showed normal foveal microstructure in both eyes (Fig. 1H and G). Work-up conducted in July 2016 for the differential diagnosis of retinal periphlebitis revealed negative anti-nuclear antibodies, p-ANCA, c-ANCA, rheumatoid factor, rapid plasma reagin, and QuantiFERON-TB Gold test. Serum angiotensin converting enzyme and lysozyme levels were within normal limits. Thorax CT was normal.

Neurosurgical evaluation included cranial MRI conducted in July 2016 which revealed an irregular lobulated pineal mass measuring 29 × 16 × 12 mm (Fig. 2A and B). Cerebrospinal fluid (CSF) analysis was negative for tumor cells and tumor markers (AFP and β-hCG). Cerebrospinal fluid opening pressure was within normal range. Further work-up performed by Endocrinology Department revealed central adrenal insufficiency, diabetes insipidus, and mild hyperprolactinemia. Endoscopic brain biopsy and histologic examination confirmed diagnosis of germ cell tumor. The patient was started on chemotherapy in August 2016 and two courses of carboplatin, etoposide, and ifosfamide treatment were followed by focal irradiation therapy. Ophthalmic examination 3 months after treatment showed complete resolution of optic disc edema and retinal periphlebitis in the right eye (Fig. 3).

3. Discussion

We report a case with pineal germinoma who presented with unilateral ocular involvement characterized by optic disc edema and retinal periphlebitis. Although a similar paraneoplastic ocular involvement was reported previously as a novel clinical entity in two separate case reports[3,4] our patient differs from those because of unilateral ocular involvement.

Paraneoplastic syndrome is characterized by distant effect of a neoplasm causing alteration of function or destruction of remote and unrelated organs by secretion of functional peptides or hormones from the tumor or inappropriate immune cross-reaction between normal host cells and tumor cells.[5,6] A novel and rare ocular paraneoplastic syndrome characterized by bilateral optic disc edema and retinal periphlebitis was reported in pineal germinoma.[3,4] Chang and associates reported on a 14-year-old boy with symptoms of increased intracranial pressure, intermittent auditory bruit, and unsteady gait and tremor. Visual acuities were 6/12 in both eyes, and the patient had diplopia and dorsal midbrain syndrome.[3] Diagnosis of pineal germinoma was established after presentation with ocular and neurological findings. Forooghian and associates reported on a 14-year-old boy who was diagnosed with pineal germinoma and who presented with 20/20 vision and dorsal midbrain syndrome.[4] Both patients had bilateral optic disc edema and retinal periphlebitis as fundus finding.[3,4] While one of the patients had bilateral vitreous cells and vitreous clumps in the inferior vitreous base[3] the second patient had a clear vitreous in both eyes.[4] Our case was a young adult who was older than the previously reported patients and was of male gender and had the same fundus findings as the two previously reported patients. However, ocular involvement was unilateral and the vitreous was clear in the involved eye in our patient. Our patient also had signs of dorsal midbrain syndrome, double vision, and endocrine abnormalities at presentation. A diagnosis of pineal germinoma was established upon brain biopsy.

Cancer associated retinopathy (CAR) is a well-recognized ocular paraneoplastic syndrome which is characterized by progressive vision loss, abnormal electroretinography (ERG), visual field deficits, and presence of circulating anti-retinal autoantibodies.[5,7–10] In CAR autoimmunity directed against retinal antigens usually leads to bilateral retinal symptoms and signs although asymmetric between the eyes.[7–9] However, there are two separate case reports showing unilateral involvement in CAR.[11,12] Roels and associates reported a case with unilateral visual symptoms consistent with CAR, abnormal unilateral ERG, and positive autoantibodies against TRPM1.[11] The patient was diagnosed with ovarian adenocarcinoma after prompt suspicion for CAR.[11] The second case by Javaid and associates presented with unilateral