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Bilateral aplasia of the optic nerve, chiasm, and tracts in an otherwise healthy infant

Optic nerve aplasia is a rare developmental anomaly characterised by the congenital absence of the optic nerve, central retinal vessels and retinal ganglion cells that is seen most often in a unilaterally malformed eye.^{1,2} Unilateral optic nerve aplasia is generally associated with otherwise normal brain development while bilateral optic nerve aplasia is usually accompanied by severe and widespread congenital central nervous system (CNS) malformations.^{1,2} We report here a unique presentation of a unilateral microphthalmos associated with bilateral optic nerve, chiasm, and tract aplasia without any CNS or systemic abnormalities.

Case report

An 11 month old girl was referred to the neuro-ophthalmology clinic because of unequal size of the eyes. She was born to non-consanguineous parents at 39 weeks gestation with a birth weight of 2750 g. The neonatal period was unremarkable. Developmental condition other than ocular findings was normal. She was the first child of her parents and there was no history of significant ocular disease in her family. Her parents' ocular examinations were normal.

Physical examination was normal, the patient's weight, length, and head circumference were normal for her age. Neurological examination was normal except for vision.

On examination she had no behavioural response to bright light and the pupils were not reactive to it. Right corneal diameter was 10 mm and left 7.5 mm. Both corneas were clear and anterior chambers were formed. Iris hypoplasia was found in both eyes. There was a congenital cataract in the left eye but the lens was clear in the right eye. On dilated fundus examination, the right eye showed absence of optic nerve and central retinal vessels (fig 1). Numerous large, round to oval, circumscribed, whitish to yellow areas of retinochoroidal depigmentation were visible in the periphery (fig 1).

B-scan echography showed a small left globe (axial length 14.80 mm), normal size right globe (axial length 22.30 mm), and absence of optic nerve on both eyes.

Figure 1 Fundus photograph of the right eye shows absence of optic disc, optic vasculature, and areas of retinochoroidal depigmentation.

Magnetic resonance imaging (MRI) of the orbits and brain disclosed marked asymmetry of globe size and bilateral absence of the optic nerve, chiasm and tract (fig 2A-D). There was only a rudimentary optic nerve behind both globes.

TORCH titres were negative and genetic evaluation including DNA analysis was unremarkable. Serum level of growth hormone, TSH, thyroxine, and glucose were all within normal limits.

Comment

Optic nerve aplasia denotes complete absence of the optic nerve and disc, retinal ganglion and nerve fibre layer, and optic nerve vessels. Histopathological examination usually demonstrates a vestigial dural sheath entering the sclera in its normal position, as well as retinal dysplasia with rosette formation.^{1,2} The pathogenesis of optic nerve aplasia remains speculative. It may be due to defective formation of the embryonal fissure, failure of the mesenchymal anlage of the hyaloid system to enter the embryonal fissure, or primary agenesis of the retinal ganglion cells.³

Bilateral optic nerve aplasia is rare, with most reported cases having major CNS malformations.^{1,2,4,5} To our knowledge there is only one previous report of bilateral optic nerve aplasia in an otherwise normal infant.⁶

Microphthalmos of the affected eye was a consistent finding in previous reports of unilateral or bilateral optic nerve aplasia,^{1,2,3,5-7} which was found only on our patient's left eye. To the best of our knowledge, this is the first case with bilateral optic nerve, chiasm, and tracts aplasia in an otherwise healthy infant with one microphthalmic and one normal size eye.

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Figure 2 (A) T1 weighted axial MRI demonstrates right normal size globe with a rudimentary optic nerve and left microphthalmic eye. (B) Axial MRI of chiasmatic section shows absence of chiasm. (C) T1 weighted axial MRI of optic tract section shows absence of tract signals. (D) T1 weighted sagittal MRI shows absence of optic chiasm.

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Uveitis and a subretinal mass in a patient with chronic granulomatous disease

Chronic granulomatous disease (CGD) is a rare disorder of phagocytic inability to produce the respiratory burst needed to kill ingested micro-organisms.¹ CGD typically presents early in life with recurrent life threatening bacterial and fungal infections and tissue granuloma formation.² Prophylaxis with systemic antibiotics, antifungals, and interferon γ have been shown to significantly reduce infections.^{1,2} Ocular involvement occurs in about 25% of CGD patients. We report the clinicopathological findings of a subretinal CGD lesion.

Case report

A 4 year old boy developed recurrent, cultured positive, cutaneous bacterial infections (*Serratia* spp). Four years later, ocular examination revealed 20/200 in the right eye and 20/50 in the left eye, bilateral moderate vitritis, retinal oedema and pigmentary clumps, multiple chorioretinal scars, and a large chorioretinal scar in the left eye. Despite transient improvement on antibiotics and systemic and periocular corticosteroids, his vision became worse with persistent vitritis and retinal exudation in the left eye. Six months later the patient was found to have an abnormal nitroblue tetrazolium test diagnostic of CGD.

Another 6 months after the diagnosis of CGD he was referred to NIH where p47^{phox} deficiency was detected.¹ Evaluation revealed 20/100 in the right eye, 20/200 in the left, mild conjunctival injection, chemosis, 1+ anterior chamber flare in the both eyes, and trace cells in the left. Posterior segment showed bilateral 2+ vitreous cells and trace haze, disc hyperaemia and oedema, perivascular sheathing, peripheral chorioretinal scars, and bone spicules in both eyes (fig 1A). Additionally, the left eye showed diffuse retinal oedema with marked macular

Figure 1 Fundus photographs showing (A) vitreous haze, mild disc oedema and hyperaemia, diffuse retinal oedema and perivascular sheathing, pigment atrophy, and marked macular lipid exudation; and (B) a subretinal mass (asterisk) in the inferotemporal region, perivascular bone spicule, and intraretinal pigment migration in the left eye.

lipid exudation and an inferotemporal subretinal mass where a large chorioretinal scar had been noted previously (fig 1B). This mass was worrying for intraocular granuloma, abscess, or neoplasm. Fluorescein angiography showed generalised disc and retinal vascular leakage, and staining of the subretinal mass, which appeared solid and homogeneous with low to moderate reflectivity on ultrasonography.

The biopsy of the intraocular mass revealed glial and vascular inflammatory granulation tissues admixed with abundant spindle cells stained positive for actin and myosin (fig 2). No granuloma or malignancy was identified. Special stains and cultures of the vitreous and biopsy were negative for bacteria and fungi. Vitreal interleukin-6 (IL-6) (141 pg/ml) and IL-8 (26 pg/ml) were elevated.

Postoperatively the patient developed a rhegmatogenous retinal detachment within a month and became blind in the left eye. The uveitis was controlled with prednisone and cyclosporin. Vision in the right eye maintained 20/80 without visual field or electroretinogram worsening during 3 years of follow up.

Comment

CGD with an incidence of 1/200 000 live births is caused by a congenital defect in phagocyte reduced nicotinamide adenine dinucleotide phosphate (NADPH) oxidase production of superoxide and related species.¹ This multi-component enzyme system is composed of cytosolic proteins (p47^{phox}, and p67^{phox}, p40^{phox}, and rac1/2) and membrane proteins (gp91^{phox} and p22^{phox} that form cytochrome b558), which assemble at membrane sites upon cell activation.³ The X linked CGD is caused by mutation in gp91^{phox} (60% of cases), while the

Figure 2 Microphotographs showing the subretinal mass that is composed of (A) glial component (asterisk), a cluster of inflammatory cells and hyalinised fibrous stroma (two asterisks) admixed with pigment clumps and small vessels (haematoxylin and eosin, original magnification, $\times 100$), (B) typical granulation tissue with proliferating capillaries and lymphoplasmacytic inflammatory cells in hyalinised stroma (haematoxylin and eosin, original magnification, $\times 400$), and (C) local proliferation of smooth muscle highlighted by positive staining for myosin (black), a marker for smooth muscle within the fibrovascular tissue (avidin-biotin-complex immunoperoxidase with anti-human myosin B antibody, original magnification $\times 200$).

autosomal recessive forms are caused by mutations in other genes.⁴ Patients with p47^{phox} deficiency (30% of cases), as in our patient, often have a milder clinical course than patients with X linked defects.

Ophthalmic manifestations of CGD include blepharoconjunctivitis, keratitis, optic neuropathy, and chorioretinal scars.⁵⁻⁷ Active chorioretinal lesions, vitreous haemorrhage, and retinal detachment are noted infrequently.⁸ Anterior segment involvement is presumably caused by inadequate control of normal flora, whereas the chorioretinal lesions are hypothesised to be the sequelae of infectious microemboli or granulomas. Pathological studies of intraocular CGD specimens have not demonstrated micro-organisms.⁹ Vitritis with active chorioretinal lesions progressing to panuveitis with an exudative retinal detachment has been reported in one CGD patient; histopathology of the eye showed choriocleral granuloma.⁸