A B S T R A C T

Aplasia of the optic nerve is an extraordinarily rare congenital anomaly that affects one or both optic nerves and is associated with the absence of the central retinal vessel and retinal ganglion cells. We report a case of unilateral optic nerve aplasia in a 4-month-old infant who was found to have left microphthalmos on routine postnatal check-up. Family history, antenatal history, and systemic evaluation were unremarkable. Magnetic resonance imaging showed absent left optic nerve with left microphthalmos. The optic chiasm was present and slightly deviated towards the right side. The remaining cerebral and ocular structures were normal.

CASE REPORT

Aplasia of the optic nerve
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Introduction
Optic nerve aplasia is a very rare congenital anomaly that is typically unilateral, and is characterised by congenital absence of the optic nerve, central retinal vessels, and retinal ganglion cells. Bilateral cases are exceedingly rare. Various ocular anomalies are associated with it.

Case report
A female infant weighing 3225 g was born to a 26-year-old G1P1 female at full term via caesarean section because the umbilical cord was around the neck. She was found to have left microphthalmos on routine postnatal check-up in December 2013 in Hong Kong, at the age of 3 months. Family history was negative for ocular or other birth defects. Her mother was a housewife, with no history of any major illness during the pregnancy and antenatal workup was unremarkable. Physical examination was unremarkable with normal development for her age.

Eye examination of the infant at 4 months revealed left microphthalmos, and a left convergent
Aplasia of the optic nerve is a rare congenital anomaly that is typically unilateral. It occurs sporadically in an otherwise healthy person without sexual or racial predilection, or any evidence of an inherited factor. Prenatal history is usually normal, but the possible influence of external factors such as episodes of viral infection in the first trimester, acetone exposure, or smoking during pregnancy cannot be excluded.

The pathogenesis of optic nerve aplasia remains unclear although it was first described 140 years ago. Scheie and Adler suggested that the defect in aplasia was failure of the mesoderm to enter the fetal fissure and provide vascularisation of the retina and nerve tissue. Weiter et al. doubted the defective mesodermal development, since the dural sheath (a mesodermal derivative) was present in the majority of their cases. Instead, they suggested that the ventral invagination of the optic vesicle causes nerve fibre misdirection and secondary atrophy. Yanoff et al. postulated a primary failure of the ganglion cell to develop and send out axons, resulting in a lack of induction of mesodermal ingrowth including a lack of retinal blood vessel development. Hotchkiss and Green agreed that failure of mesodermal induction was secondary to third-order neuronal defect in the ganglion cell layer.

Plain X-ray can demonstrate a small optic foramen on the side of aplasia. Computed tomographic scan may show the globe and orbit on the affected side to be smaller than the normal side. Magnetic resonance imaging will show the absence of optic nerve on the affected side. The chiasm and lateral geniculate body may also appear small.

Histopathological findings in optic nerve aplasia include the absence of ganglion cells and their axons as well as the absence of retinal vessels.

According to many previous statements, aplasia of the optic nerve is a part of hypoplasia of the optic spectrum. According to an analysis performed by Alqahtani, of 42 cases in the literature, 29 were genuine aplasia of the optic nerve, while the remainder were hypoplastic optic nerve.

Unilateral aplasia of the optic nerve is often present in malformed eyes, with no abnormality in brain tissue. Possible malformations of the eye include microphthalmos, cataract, retinal dysplasia, coloboma of the iris and ciliary body, iris hypoplasia, malformation of the chamber angle, and persistent hyperplastic primary vitreous. No light perception is present in the affected eye and light stimulation...
elicits no direct or consensual pupillary response. Light stimulation of the normal eye results in a direct or consensual pupillary response.\textsuperscript{11} Possible malformation of the central nervous system includes hydranencephaly, orbital meningoencephalocele, and anencephaly.\textsuperscript{5,6}

The prognosis of optic nerve aplasia is poor. There is no specific treatment and blindness occurs in the affected eye. Management of such cases is directed towards identifying any associated ophthalmological or neurological problems. Cavallini et al\textsuperscript{12} recommended ocular prosthesis in patients with associated microphthalmos, to enable normal development of the orbit, at least for aesthetic purposes.

Optic nerve aplasia should be suspected in a patient who presents with unilateral microphthalmos that is associated with the absence of central retinal vessels and ganglion cells. Magnetic resonance imaging is useful to screen for other associated intracranial abnormality.

References