Adult-onset Dandy–Walker syndrome with atypical ocular manifestations

Dandy–Walker malformation (DWM) consists of a spectrum of disorders of neural development, with a classical triad of complete or partial agenesis of the vermis; enlargement of the posterior fossa with upward displacement of the tentorium, transverse sinus, and torcular; and cystic dilation of the fourth ventricle. It is the most common human cerebellar malformation and may be associated with agenesis of corpus callosum or nonneural anomalies such as skeletal deformities, congenital heart disease, polycystic kidneys, or facial clefts.[1,2] We report the simultaneous occurrence of myopia, cataracts, and visual field defects in an adult patient with DWM.

A 19-year-old male was referred to our tertiary eye care facility for evaluation of bilateral cataracts with a history of having undergone laser in-situ keratomilieusis elsewhere for axial myopia of −7.00 Dsph in both eyes about 3 years ago.

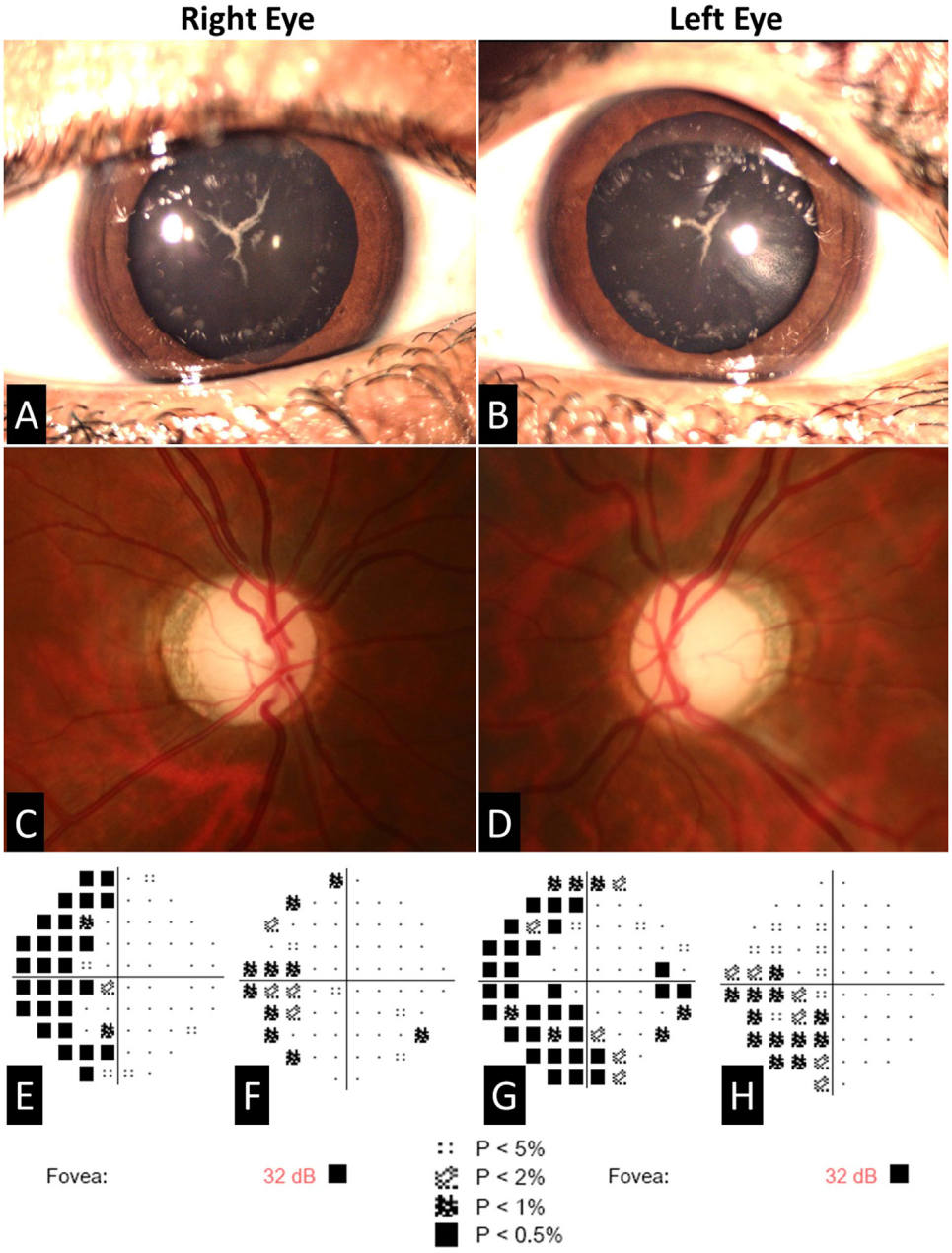

Fig. 1—Ocular findings in the index case included bilateral sutural and blue-dot cataracts (A, B), myopic temporal and nasal crescents and mild temporal pallor in both optic discs (C, D), left homonymous hemianopia on Humphrey's 30-2 visual field testing (E, G), right macular sparing (F), and left inferotemporal quadrantic macular field defect (H) on 10-2 visual field testing.

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No physical records from that surgery were available for review. Ocular examination showed unaided visual acuity of 6/7.5 in both eyes, normal ocular motility, well-apposed laser in-situ keratomilieusis flaps with no interface debris, bilateral sutural and blue-dot cataracts (Fig. 1A, B), myopic fundi with no peripheral lesions, and temporal pallor of both optic discs (Fig. 1C, D). Axial lengths were 25.76 mm, as measured on optical biometry of both eyes. Ductions, versions, vergence, saccades, and pursuit movements were within normal limits. There was mild nystagmus in extremes of dextro- and levoversion. Automated perimetry (Humphrey's 30-2) of both eyes showed left-sided homonymous hemianopia with relative sparing of the central visual fields (Fig. 1E, G). Humphrey's 10-2 testing showed a relatively spared right macula and a left inferotemporal quadrantic macular field defect (Fig. 1F, H). The foveal thresholds were 32 dB in both eyes ($p < 0.5$). On further questioning, the patient revealed occasional difficulty in walking with a history of falling down twice in the past 6 months. Suspecting a neurologic cause of the visual field defects, non-contrast-enhanced computed tomography of the head was done, which showed ventriculomegaly, moderate hydrocephalus, mega cisterna magna, and high attachment of the tentorium cerebelli suggestive of DWM (Fig. 2A).

The patient was referred to a neurosurgeon and followed up biannually. The visual field defects were found to be non-progressive. The patient underwent uneventful ventriculoperitoneal shunting 2 years later due to worsening gait instability and weakness (Fig. 2B). In response to the moderate hydrocephalus that was noticed on preoperative radiologic scan, the patient was premedicated with oral acetazolamide and intravenous mannitol but did not respond. Intraoperatively, the ventricle was punctured by a ventricular needle, and the cerebrospinal fluid pressure was measured to be >20 mm Hg (raised). Thus a medium-pressure ventriculoperitoneal shunt was implanted. The patient had stable (slightly weaker than normal) gait until his last follow-up.

Adult-onset of the DWM presenting with ocular manifestations has been reported rarely because the condition usually presents as a congenital defect with hydrocephalus or cerebellar signs and may even be asymptomatic at presentation.[3–7] The ophthalmic associations of DWM have not been widely studied except as part of the PHACES syndrome (posterior fossa anomalies, infantile hemangioma of the head or neck, cardiac defects, eye abnormalities, and sternal or ventral defects).[3,7] Isolated reports of findings include myopia, microcorneas, sclerocornea, posterior embryotoxon, shallow anterior chamber, iris depigmentation, persistent pupillary membrane, and ocular colobomas (Aicardi syndrome). Fundus abnormalities include retinal dysplasia, nonrhegmatogenous retinal detachment, foveal hypoplasia, macular edema, nummular macular depigmentation, retinitis pigmentosa (Usher syndrome), and persistent hyperplastic primary vitreous (Warburg syndrome). Tearing while chewing (cervico-oculo-acoustic syndrome) and aberrant ocular motility in the form of nystagmus, bilateral Duane's retraction syndrome, Joubert–Boltshauser syndrome, and Moebius syndrome are the neuro-ophthalmic manifestations reported. There are few reports of the association of DWM with cataracts,[3] and the occurrence of bilateral sutural or blue-dot cataracts as a manifestation of DWM presenting in adulthood is exceedingly rare. In addition, our patient had temporal pallor of the optic discs, which is likely to be missed in high-myopic patients. Subsequent perimetry revealed visual field defects pointing to an occipital lobe lesion arising out of the anatomic malformation, which, corroborated by the retrospectively acquired history of gait instability, was diagnosed as DWM on a subsequent computed tomographic scan. The case could have benefitted from additional molecular analysis of genes associated with DWM, but because of lack of access to testing facilities, this was not possible and is a limitation of this case report. However, a high index of suspicion for syndromic associations of juvenile cataract and meticulous neuro-ophthalmic work-up including neuroimaging led to early diagnosis and appropriate management of the condition in our patient.

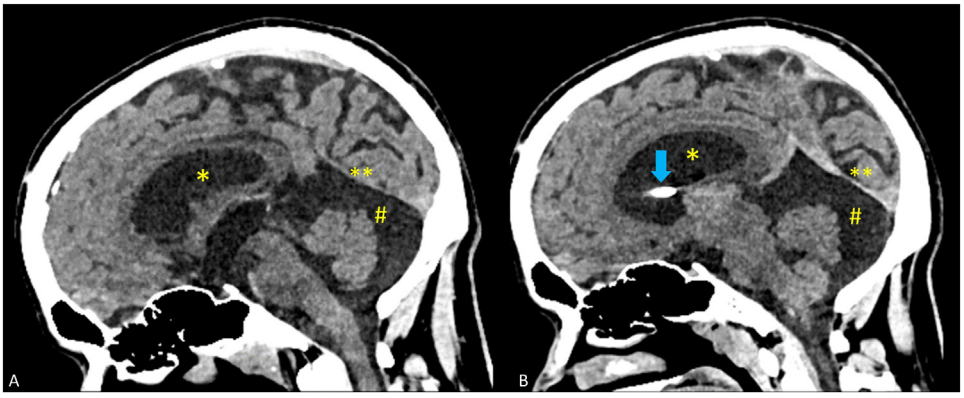

Fig. 2—Non-contrast-enhanced computed tomography of head preoperatively (A) showing normal calvarium, ventriculomegaly with periventricular ooze (*), mega cisterna magna (#), and high attachment of the tentorium cerebelli (**) suggestive of Dandy–Walker malformation. (B) Postoperatively, ventricular drain tip is seen in the anterior body of the left lateral ventricle (blue arrow).

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Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this correspondence.