Bilateral macular colobomata: Temporal dragging of optic disc

David J Mathew

A 13-year-old male presented with decreased vision and squint from childhood. He had bilateral large colobomata at the macula in each eye, the one on the right being larger than the left. The disc was dragged temporally with straightening of the temporal retinal vessels. This is a case report of bilateral large macular coloboma and serves to report its association with a temporally dragged disc and straightened temporal retinal vessels. A dragged disc if present with a colobomatous defect at the macula may strengthen the case for diagnosis of macular coloboma and help exclude other differentials.

Key words: Dragged disc, macular coloboma, macular coloboma differentials

Congenital macular colobomata are well-circumscribed, punched out atrophic lesions of varying size, usually solitary, located at the macula. These resemble other conditions, namely congenital toxoplasmosis macular scar, North Carolina macular dystrophy, Leber's congenital amaurosis, advanced cone dystrophy, and central areolar choroidal dystrophy. This report is a case of bilateral large macular colobomata with the observation of signs hitherto unreported in literature to the best of my knowledge.

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References
Case Report

A 13-year-old male presented with a history of decreased vision in both eyes and squint from childhood. He reportedly had a fall from the bed at 5 months of age without any obvious consequences. There was no history of maternal fever, significant maternal antenatal history or complications during birth. There was no significant family history.

The best-corrected visual acuity was 20/120 OD and 20/80 OS as measured by the Snellen’s visual acuity chart. He was trichromatic in both eyes. He had a chin down head posture with right exotropia. Right eye suppression was detected using Worth’s four dot test. Fundus examination in each eye revealed a large, well-demarcated excavation at the macula, spanning both the temporal arcades with baring of the underlying sclera and a thin rim of pigmentation at the periphery, consistent with a coloboma [Fig. 1]. The coloboma was larger on the right side. Temporal dragging of the optic disc and straightening of the temporal arcade vessels, thus reducing the angle between them, were present. The nasal arcade vessels appeared to have a proximal temporal loop before turning nasally, a finding noted commonly with dragged discs [Fig. 2]. Rest of the retina was within normal limits.

As he was a school student, low vision aids were given to help him see better in the classroom.

Discussion

Macular colobomata are believed to be a consequence of arcuate bundles along the horizontal raphe undergoing incomplete differentiation.[1] As such, these are atypical colobomata as they do not fall in areas that originate from the embryonic cleft. Other similar lesions include congenital toxoplasmosis macular scar, North Carolina macular dystrophy, Leber’s congenital amaurosis, advanced cone dystrophy, and central areolar choroidal dystrophy.

Toxoplasma retinochoroiditis is usually unilateral. There may be reactivation with the average number of attacks being 2.7/patient.[2] Though sharply demarcated, the edges of these lesions are not well-rounded, or oval shaped like a coloboma. Serological confirmation can be obtained in warranted cases.

North Carolina macular dystrophy is autosomal dominantly inherited with complete penetrance. Grade 3 of this entity resembles macular coloboma.[3]

Leber’s congenital amaurosis presents with blindness and nystagmus or roving eye movements. Pigmentary retinopathy, arteriolar attenuation and optic atrophy usually develop over time. Severe pigmentation or a coloboma-like defect may be seen in the macula. Eye poking and enophthalmos may be seen in some.[4,5]

Progressive cone dystrophy and central areolar choroidal dystrophy present between the second and fourth decades of life. The eventual lesion at the macula is geographic atrophy.[6,7]

Temporal dragging of the disc with associated vascular changes is commonly associated with retinopathy of prematurity, familial exudative vitreoretinopathy, peripheral toxocara granuloma, combined hamartoma of the retina and retinal pigment epithelium, incontinentia pigmenti, and congenital retinal folds.[8] A dragged disc with straightening of vessels if seen with a large macular coloboma as in this case can aid in the clinical diagnosis and exclusion of other differentials.

In summary, this is a case report of a 13-year-old male with bilateral large colobomata at the macula. A dragged disc with straightening of retinal vessels has been hitherto unreported in association with a macular coloboma though it may have been observed in the past. These clinical findings can aid in the diagnosis of macular coloboma and exclusion of other differentials.

References

Population estimated between 0.13% and 0.42% in healthy pseudomembranous conjunctivitis. Juvenile colloid milia, ligneous conjunctivitis, and palmoplantar keratoderma are associated with systemic pseudomembranous lesions of the skin, oral mucosa, gingiva, ears, tracheobronchial tract, female genital tract, and palms. The patient presented with redness, watering since 8 months. Her vision was 20/30 in right eye and 20/70 in left eye. Her hyperpigmented papulopustular lesions over face, palms, and legs. She was started with topical moxifloxacin, lubricating drops. Patient was followed-up after the excision of membrane, hemostasis was achieved and supratarsal injection of triamcinolone acetate was administered.

The conjunctiva had plenty of purulent discharge and superficial punctate keratitis, epithelial defects, and peripheral vascularization. Anterior chamber was quiet. Pupils were normal in size and reacting to light. Fundus appeared normal in both eyes. There was no history of fever, instillation or intake of any drugs into the eyes. Anterior segment examination was normal. Slit lamp examination showed matting of lashes with crusted discharge. Conjunctiva had plenty of purulent discharge and conjunctival hyperemia. There was no underlying cicatricial conjunctivitis, tarsal conjunctivitis, and watery discharge. There was no history of tuberculosis. Patient was followed-up after the excision of membrane, hemostasis was achieved and supratarsal injection of triamcinolone acetate was administered.

Ocular and extra-ocular features of patients with Leber congenital amaurosis were normal. It may be associated with systemic pseudomembranous lesions of the skin, oral mucosa, gingiva, ears, tracheobronchial tract, female genital tract, and palms. The patient presented with redness, watering since 8 months. Her vision was 20/30 in right eye and 20/70 in left eye. Her hyperpigmented papulopustular lesions over face, palms, and legs. She was started with topical moxifloxacin, lubricating drops. Patient was followed-up after the excision of membrane, hemostasis was achieved and supratarsal injection of triamcinolone acetate was administered.

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