The risks for ocular toxoplasmosis, both primary and recurrent disease, are still largely unknown. In the immunocompetent host, clinical features of ocular toxoplasmosis disease are related to patient age; patients in their second through fourth decades of life account for most observed episodes.[7,8] It is unknown in our patient whether *T. gondii* infection was a congenital infection or postnatal acquisition. Testing of the mother was never obtained; PCR testing of the CSF was obtained when the child was hospitalized.

HIV/AIDS has provided evidence that host defenses are important for limiting the severity of ocular toxoplasmosis. Patients with immunosuppression, resulting from congenital causes as well as in those receiving immunosuppressive drug therapies, may be at similar risk as a patient with HIV/AIDS. Host changes in lymphocytes, natural killer cells, macrophages, and cytokine production, all known to be involved in host defenses against *T. gondii* may predispose to disease.[8,9] Central nervous system infection with *T. gondii*, apart from the eye, occurs exclusively in immunocompromised individuals. *T. gondii* is the most common cause of brain abscess in immunocompromised hosts.

Despite normal measured IgG and CD4+ levels, our patient developed disseminated toxoplasmosis infection. Hospitalization for disseminated *T. gondii* infection might have been prevented had prophylaxis been considered sooner.[10] There are no previous reports of toxoplasmosis/opportunistic infection in a patient with AT, making it difficult to consider recommendations for prophylaxis. It is reasonable to recommend children diagnosed with AT or other immune-compromising diseases be followed by an ophthalmologist.

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Upbeat nystagmus in a 3.5-year-old boy

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Upbeat nystagmus is usually a central vestibular nystagmus attributable to structural brainstem or cerebellar lesions. Adult-onset upbeat nystagmus calls for a thorough neurological evaluation. In children, however, it can occur as a transient phenomenon in healthy neonates or as a sensory form of nystagmus that usually transforms into horizontal nystagmus by 2 years of age. We report the case of 3.5-year-old boy with upbeat nystagmus. His ocular examination was within normal limits. Neurological evaluation was normal. Optical coherence tomography testing and Electroretinogram confirmed cone dysfunction. Over the next 6 months the upbeat nystagmus converted to horizontal nystagmus.

Case Report

A 3.5-year-old boy child presented at Narayana Nethralaya-2, Bengaluru, with shaking of the eyes and aversion to bright light since 7 months of age. He was a first child, born of second-degree consanguinity, with normal birth and developmental history. Family history was noncontributory. On examination, his unaided visual acuity was 20/360 in each eye with Teller acuity cards at 55 cm, with 75% reliability. He maintained a mild chin-up posture during examination and was photophobic throughout. He had an upbeat nystagmus of high frequency and low amplitude in primary position and down-gaze but increased in frequency in upgaze (Video 1, available at jaapos.org). Cover testing was unremarkable. Ductions and versions were full. Examination of the anterior segment and fundus (Figure 1) were within normal limits. Cycloplegic refraction showed regular astigmatism in both eyes. Systemic examination revealed no abnormality. Persistence of upbeat nystagmus beyond 3.5 years in absence of any structural ocular feature prompted magnetic resonance imaging of the brain; results of imaging and neurological examination were normal.

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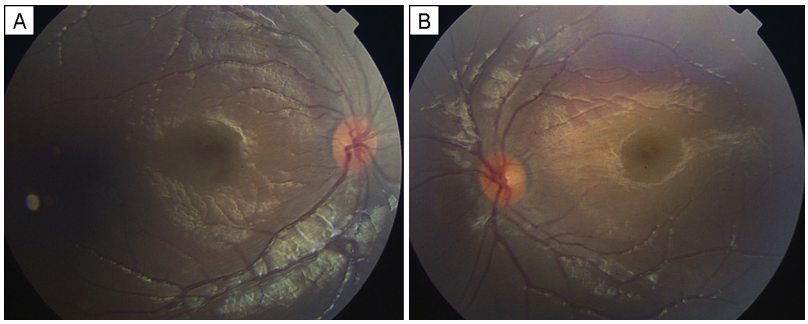

FIG 1. Fundus photographs showing normal disk and macula, with normal vasculature, in the right eye (A) and left eye (B).

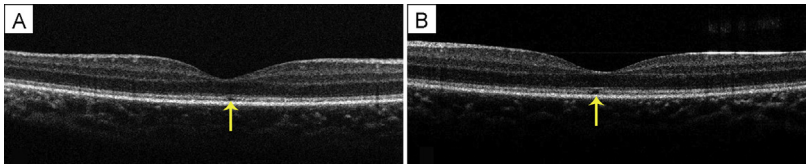

FIG 2. Spectral domain optical coherence tomography (Envisu, Bioptigen, NC) of the right eye (A) and left eye (B) showing an attenuated inner segment–outer segment layer, with an absent outer segment–retinal pigment epithelium layer with an absent foveal tent (arrow) in the foveal center, denoting loss of foveal cones.

In view of the photophobia, history of consanguinity, normal ocular anatomy, and normal neurological findings, an optical coherence tomography (OCT) was advised. Handheld spectral domain SD-OCT (Envisu 2300; Bioptigen Inc, Research Triangle Park, NC) was performed and showed an attenuated inner segment–outer segment (IS-OS) layer, absence of an outer segment–retinal pigment epithelium layer, and absence of a foveal tent in the foveal center, denoting loss of foveal cones in both eyes (Figure 2). A full-field electroretinogram (ERG) was performed with a handheld stimulator (Monbaby Stimulator; Metrovision, Perenchies, France) under general anesthesia. The ERG revealed normal a- and b-waves on scotopic flashes to 0.01 cd/m^2 and 3.0 cd/m^2 flashes, with no recordable response to the photopic 3.0 cd/m^{2}flash stimulus (e-Supplement 1, available at jaapos.org), with the 30 Hz 3.0 cd/m^2 flicker response showing an unreliable response, because it was contaminated with electrical noise during testing. Based on these findings, the patient was diagnosed with complete rod monochromatism (or achromatopsia), with a differential diagnosis of a cone dystrophy. He was prescribed photochromatic glasses and a hooded cap to avoid photoaversion. At follow-up examination 8 months later his visual acuity had not deteriorated further in both eyes, confirming the vision stability expected in rod monochromatism. His nystagmus in the primary position showed a change from an upbeat to a very low-amplitude and very low-frequency horizontal nystagmus, like a shimmering movement (Video 2, available at jaapos.org).

Discussion

Common etiologies for anterior visual pathway diseases associated with sensory nystagmus include Leber congenital amaurosis, congenital retinal dystrophies, congenital albinism, and optic atrophy. Onset of nystagmus typically occurs by 2-6 months of age. Clinically, sensory nystagmus is characterized by a pendular or jerky nystagmus, which is usually horizontal or rotatory. It remains horizontal in upgaze and can worsen with attempted fixation or intense visual effort. It is not associated with oscillopsia.[1] Occasionally it can be vertical, like upbeat nystagmus.

Upbeat nystagmus has been described as a transient phenomenon in healthy neonates and has been attributed to immature central vestibular connections associated with vertical canal inputs.[2] Spontaneous upbeat nystagmus due to focal central lesions results from a primary dysfunction of the superior vestibular nucleus ventral tegmental tract pathway, which becomes hypoactive after pontine or caudal medullary lesions.[2] A positive family history and normal visual acuity may indicate a familial upbeating nystagmus.[1] In their study of 131 infants with congenital nystagmus, Hoyt and Gelbart[3] found 9 cases of vertical nystagmus associated with congenital ocular abnormalities; 4 infants had upbeat nystagmus, which became horizontal in 3 infants within 1 year of age. The patient with persisting upbeat nystagmus had cerebellar vermis hypoplasia. Good and colleagues[4] studied 11 children with upbeat nystagmus in infancy due to anterior visual pathway diseases that became a horizontal nystagmus in the first 2 years of life. Shawkatet and colleagues[5] studied 276 children with infantile nystagmus; of these, 11 children with vertical nystagmus had either a sensory defect nystagmus or congenital idiopathic nystagmus. Of these 11, 7 had congenital cone dysfunction, 1 had cone-rod dystrophy, 1 had congenital stationary night blindness, and 2 were diagnosed as congenital idiopathic nystagmus, confirmed on

ERG and visual evoked potential testing. The vertical nystagmus in their study persisted into childhood, although the time frame was not mentioned. Our case had an upbeat nystagmus that persisted beyond 3.5 years of age, similar to the study of Shawkat and colleagues, but eventually transformed into a horizontal nystagmus. This late transformation has not been reported previously.

SD-OCT alone cannot diagnose cone-rod dystrophies; however, it is emerging as a useful adjunctive imaging tool for better characterization of lesions.[6] Using handheld OCT, Yang and colleagues[7] measured the relative intensity of the foveal ellipsoid zone in children with achromatopsia: in 67% of patients it appeared disrupted; in 2, it appeared intact but lacked the prominent foveolar outer segment thickening; and in 1 it appeared normal.

Hood and collagues[8] showed that in patients with cone dystrophy the intensity of IS-OS junction band seen on SD-OCT was lower in patients with diminished cone function than in healthy controls. Sergouniotis and colleagues[9] reported disruption of IS-OS junction in 12 patients with cone dystrophy. Our patient also showed disrupted IS-OS junction and loss of foveal tent. In cone dystrophy, cone function is normal at birth and typical symptoms (reduced visual acuity, photophobia, increased sensitivity to glare, and abnormal color vision) appear later.[10]

Differentiating between achromatopsia and cone dystrophy can be difficult, particularly in individuals with onset in early childhood; the best clinical discriminator is disease progression, which occurs in cone dystrophy and but typically not in individuals with achromatopsia.[10] Because the visual acuity in our case did not worsen during the follow-up period, achromatopsia is the more likely diagnosis.

In conclusion, children presenting with vertical nystagmus should undergo a detailed retinal check-up with OCT as the first line of investigation. SD-OCT can help us categorize retinal dystrophies, which can be confirmed by ERG. Neuroimaging can be reserved for cases in which ERG is found to be normal, because neuroimaging is expensive and requires that the child be under anesthesia for the procedure and this sensory type of vertical nystagmus can persist later into childhood before developing into a horizontal type.

Literature Search

PubMed was searched on April 2, 2015, without language or date restriction, using the following terms: *upbeat nystagmus*, *achromatopsia*, and *OCT imaging*.

Acknowledgments

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