Optic Nerve Appearance in Patients With Down Syndrome

To the Editors:

Children with Down syndrome (DS) are commonly seen by pediatric ophthalmologists because significant ocular conditions are associated with the disorder.1 The presence of optic nerve anomalies has been noted in various small case series on children with DS.2,3 We sought to determine the prevalence and character of optic nerve abnormalities among patients with DS in a large pediatric cohort and to ascertain whether patients with DS and optic nerve findings suggestive of an intracranial process (nerve elevation or pallor) had corresponding central nervous system pathology.

A retrospective review of the charts of all patients with DS (10-year study period) seen by the ophthalmology service at Boston Children’s Hospital was performed to identify those with optic nerve anomalies. One hundred sixteen patients with DS and optic nerve abnormalities were identified (116 of 806, 14% of patients with DS). The most frequently identified abnormality was a myopic appearance and/or peripapillary atrophy of the optic nerve head. Other anomalies included peripapillary pigmentation, scleral crescent, optic nerve pallor, and non-myopic peripapillary atrophy (Figure 1).

Seventeen patients had elevated optic nerve heads (17 of 806, 2%), eight of whom underwent neuroimaging. No imaged patient was found to have pathology compatible with optic nerve swelling. B-scan ultrasonography was performed on five patients, either after neuroimaging or as a primary diagnostic measure. Optic nerve head drusen were found in all of these children. Four patients with DS in the cohort had pale optic nerves. Optic nerve head anomalies were not attributable to an undiagnosed intracranial lesion in any case.

Among the optic nerve anomalies identified in our cohort, we focused particularly on those that could be associated with a vision-threatening or life-threatening process: optic nerve head elevation and optic nerve pallor. In previous small case series, optic nerve head elevation was noted in 3% to 5% of patients with DS.2,4

In this investigation, none of those with elevated or pale nerves had undiagnosed life or vision-threatening pathology either clinically or by neuroimaging. This is consistent with prior research on CNS lesions in the DS population because solid intracranial tumors appear to be extremely rare in these patients.5

Based on our findings in this study, we recommend a systematic approach to evaluating children with DS who have elevated-appearing optic nerves. B-scan ultrasonography may be employed as a first-line diagnostic modality in these patients. It is inexpensive, non-invasive, and sensitive for optic disc drusen. A positive scan obviates the need for costly and invasive testing because drusen can be monitored ophthalmoscopically and with visual field testing. In the case of a negative scan, further measures should be considered, including neurologic consultation, neuroimaging, and lumbar puncture. Patients who demonstrate nerve pallor of unknown etiology may prompt neuroimaging in addition to neurologic and/or neuroophthalmic consultation.

REFERENCES


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Retrobulbar Optic Neuritis and Live Attenuated Influenza Vaccine

To the Editors:

Retrobulbar optic neuritis is rare in childhood and even rarer when associated with vaccinations. Select case reports have shown association with vaccination against hepatitis B, influenza, tetanus, measles, and rubella, yet it has not been cited following a nasal influenza vaccination. We present evidence for a causal relationship between optic neuritis and the nasally administered live attenuated influenza vaccine.

A 13-year-old boy developed severe vision loss in both eyes 2 weeks following a nasal influenza vaccination. He presented with best-corrected visual acuity of 20/counting fingers at 6 feet in the right eye and 20/light perception in the left eye. A positive right relative afferent pupillary defect was noted. Extraocular movements were full with mild guarding at extremities of gaze. Anterior segment and posterior segment examination was unremarkable. There was no papillitis or papilledema. A global visual field defect was present in both eyes, based on Humphrey 30-2. Magnetic resonance imaging showed diffuse enlargement of the optic chiasm with inflammation of distal optic nerves bilaterally. His medical history was only significant for asthma and childhood obesity and there was no prior history of multiple sclerosis, cat bites, tick bites, or autoimmune diseases.

Two weeks prior, he received the nasal influenza vaccine, a trivalent vaccine consisting of three influenza viruses: A(H3N2), A(H1N1), and B virus.

Full blood panel testing to include infectious and inflammatory etiologies was normal. Cerebrospinal fluid analysis was normal. With the exception of optic chiasm enlargement, magnetic resonance imaging showed no additional white matter lesions separated in space or time.

Our patient received intravenous methylprednisolone 30 mg/kg/dose over a 30-minute period once daily for 3 days. Visual acuity improved from light perception in both eyes on hospital day 2 to counting fingers at 3 feet in both eyes on the day of discharge. The patient underwent a 3-week course of tapering oral prednisone. Three months after the initial incident, his visual acuity drastically improved to 20/20 in both eyes and his visual field returned to normal with no residual visual field defect.

Nasal influenza vaccination has been available to the public since 2006. It is implemented in numerous school-wide vaccination campaigns. Its ease of use and administration are two desirable advantages over the traditional intramuscular influenza vaccine. One concern of nasal influenza vaccination is related to its mode of delivery proximity to the brain. Theoretically, live vaccine particles have direct access to the brain after the cribiform plate located at the apex of the nasal sinuses. This theory may explain our patient’s uncommon presentation of diffuse inflammation of the optic chiasm causing bilateral optic neuritis.

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