Neuroimaging Findings in Patients With Down Syndrome and Nystagmus

To the Editors:

Children with Down syndrome reportedly have a high prevalence of nystagmus.1-3 The etiology of nystagmus in Down syndrome has been presumed to be idiopathic, although evidence for this is limited. Whether cerebellar hypoplasia, a common magnetic resonance imaging (MRI) finding in Down syndrome, contributes to nystagmus has been investigated recently because of the role of the cerebellum on gaze-holding mechanisms within the brain.4 We sought to report the prevalence of nystagmus in a large cohort of patients with Down syndrome and to determine whether underlying progressive neurologic disease contributes to nystagmus.

We performed a retrospective chart review of all patients with Down syndrome seen in the Ophthalmology Department at Boston Children's Hospital from 2000 to 2010 who were diagnosed as having nystagmus on clinical examination. Patients with manifest nystagmus, latent nystagmus, and manifest latent nystagmus were included. Neuroimaging results for computed tomography (CT) and MRI were recorded when performed, and the primary indications for neuroimaging were reviewed.

A total of 806 patients with Down syndrome were identified. The prevalence of nystagmus was 17% (138 of 806), and nystagmus was binocular in all but one case (137 of 138). Manifest nystagmus occurred in 59% (81 of 138), latent nystagmus in 9% (12 of 138), and manifest latent nystagmus in 32% (45 of 138) of patients with Down syndrome.

Neuroimaging was performed specifically for the indication of nystagmus in 5% (7 of 138) of patients. Results were completely normal in 4 patients. In 2 patients, unrelated, benign findings of choroid plexus cysts and asymmetric enlargement of the lateral ventricles were described. In 1 patient, dysmorphic cerebellar hemispheres were noted. With the exception of the cerebellar findings, no other results were potentially related to the etiology of the nystagmus.

Neuroimaging was performed in 19 additional patients with Down syndrome for other neurologic indications, including but not limited to a history of infantile spasms, headache, vertigo, and developmental delay.

In total, 19% (26 of 138) of patients with Down syndrome with a clinical diagnosis of nystagmus had neuroimaging, and in no case was a progressive neurologic process found to explain the presence of nystagmus. Three of these children were noted to have cerebellar hypoplasia.

Other studies have shown rates of nystagmus in patients with Down syndrome similar to our study's prevalence of 17%. For example, da Cunha and Moreira5 demonstrated 18% prevalence in a cohort of 152 children with Down syndrome, and Stephen et al.6 described 16% prevalence in a cohort of 72 patients. To date, we report the prevalence of nystagmus in the largest cohort of pediatric patients with Down syndrome of which we are aware.

Nystagmus in pediatric patients with Down syndrome can prompt the question as to whether or not imaging is warranted. Neuroimaging is not entirely without risk in this population. There are risks related to sedation, side effects of dye used to enhance imaging, and potential exposure to radiation with CT. In our cohort, neuroimaging did not reveal any underlying progressive neurologic process as the basis for nystagmus. In the absence of other focal neurologic signs or indications, neuroimaging to exclude progressive neurologic disease as a cause of nystagmus may be of low yield in the population with Down syndrome.

REFERENCES


Melissa M. Wong, MD
A.J. Schneier, MD
Danielle Ledoux, MD
Gena Heidary, MD, PhD
Boston, Massachusetts

The authors have no financial or proprietary interest in the materials presented herein.
doi:10.3928/01913913-20161021-02