JAMA Ophthalmology Clinical Challenge

Abnormal Eye Movements in a Young Girl

Samantha J. Sechrist, BS; Christine M. Glastonbury, MD; Alejandra G. de Alba Campomanes, MD

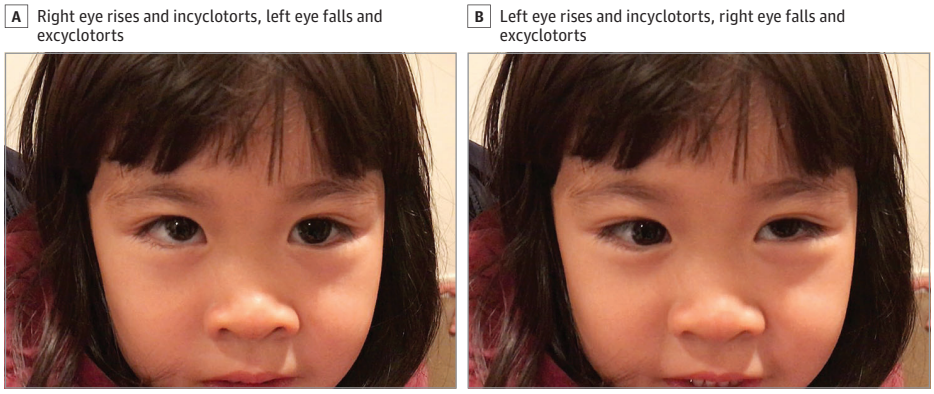

Figure 1. Seesaw nystagmus shown in 2 half cycles.

A 5-year-old girl was referred to ophthalmology care for evaluation of nystagmus. Her medical history included esophageal atresia, small bowel obstruction, mild cognitive impairment, low tone, and increased reflexes. Prenatal history was complicated by insulin-controlled gestational diabetes and polyhydramnios. Her ocular history was notable for eye movement abnormalities since birth. She had no family history of ocular disease.

On examination, her visual acuity was 20/150 OU and did not improve with correction (right eye, −1.50 [2.00] × 180; left eye, −1.00 [1.50] × 180). Pupillary responses were normal. Alternating vertical and torsional nystagmus was noted (**Figure 1**). Specifically, 1 eye was noted to rise and intort while the other would fall and extort, followed by reversal of the vertical and torsional components in the opposite eye (**Video**). The optic nerve and foveal reflexes were normal in both eyes. Flash visual evoked potential (VEP) demonstrated strong ipsilateral activity of the occipital lobe. Results of a previously obtained brain magnetic resonance imaging (MRI) scan were reported as normal.

WHAT WOULD YOU DO NEXT?

A. Review brain MRI and order electroretinogram

B. Refer for strabismus surgery

C. Order genetic workup

D. Treat with clonazepam

CME Quiz at jamacmelookup.com

Multimedia

Diagnosis

Achiasma with seesaw nystagmus

What to Do Next

A. Review brain MRI and order electroretinogram

Discussion

Seesaw nystagmus (SSN) is a rare pendular nystagmus characterized by elevation and intorsion of 1 eye and synchronous depression and extorsion of the other eye for a half cycle with reversal of these movements constituting the second half cycle to form seesawlike movements, as seen in this patient. Etiologies can include achiasma, septo-optic dysplasia,[1] albinism, retinal dystrophies, and Chiari malformation.[2] Acquired forms can result from pituitary adenoma, head trauma, brain stem infarction, whole brain radiation, and intrathecal methotrexate.[2] Achiasma is the absence of the optic chiasm when nasal optic nerve fibers fail to decussate to the contralateral hemisphere and is associated with nystagmus, strabismus, and decreased visual acuity.[1] Diagnosis is made by absent optic chiasm and nonsegregation of the visual pathway on MRI.

Choice A, review brain MRI and order an electroretinogram (ERG), is the correct next step. MRI reads may miss achiasma, especially if thick sections are evaluated or if the optic pathway is not carefully assessed.[1] The VEP in this patient showed the right eye projecting to the right ocular cortex and the left eye to the left cortex (thus each cortex was receiving complete but monocular visual field). Therefore, achiasma should be suspected despite the initially normal MRI results. These VEP findings contrast with albinism, in which

VEP shows interocular contralateral asymmetry.[3] An ERG is useful to rule out SSN due to retinal dystrophies. Regardless of waveform, in the workup of infantile nystagmus, an ERG is of high diagnostic yield, whereas MRI is more useful when neurological abnormalities or an unusual nystagmus pattern is present.[4,5] This case illustrates that an MRI scan may be erroneously interpreted as normal if the imaging protocol and assessment are not specific for the suspected ophthalmic pathology.

Strabismus surgery (choice B) is incorrect as surgery for SSN is not well established.[6] Genetic testing can be an important step in the general workup of nystagmus in children[5]; however, the genetic mechanisms of achiasma are not understood,[7] therefore, genetic workup (choice C) would not guide management. Achiasma can be associated with craniofacial defects, heart defects, agenesis of the corpus callosum, and vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, kidney anomalies, and limb abnormalities (VACTERL) syndrome.[8] Similar to this patient, an association with esophageal atresia has been reported.[9] Clonazepam may be effective in acquired SSN,[10] but there is a lack of evidence for congenital SSN; therefore, option D is incorrect.

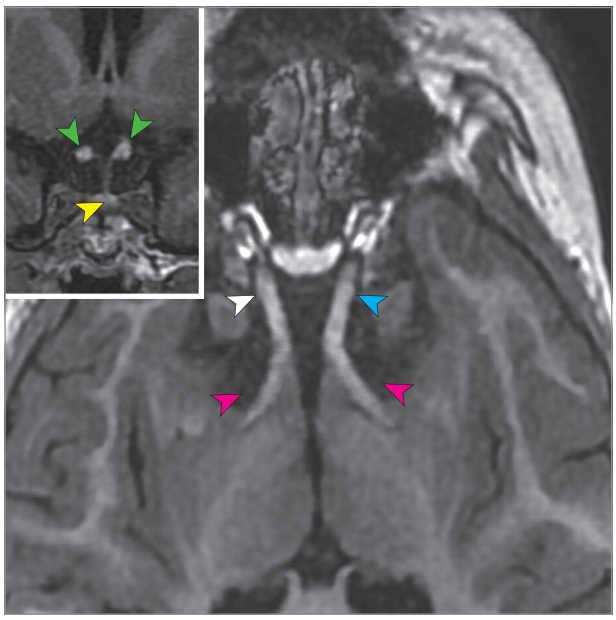

Figure 2. Oblique axial image from 3-dimensional spoiled gradient recalled echo (SPGR) sequence along the right (white arrowhead) and left (blue arrowhead) optic nerves shows no meeting of the nerves to form a chiasm, but each nerve coursing laterally to form optic tracts (pink arrowheads). Inset shows coronal image from the same 3-dimensional SPGR sequence at level of pituitary stalk and gland (yellow arrowhead) shows separate white matter bundles (green arrowheads) where a chiasm is normally evident.

Patient Outcome

A second read with guidance regarding clinical suspicion of achiasma was ordered and found normal contour, caliber, and signal intensity of the optic nerves with a normal course into the suprasellar cistern, but no chiasm was identified. The optic tracts deviated in the expected lateral direction and superolateral plane without neural or fibrous connections between the optic nerves (**Figure 2**). When suspecting achiasma, 3-T imaging with thin-slice T2 fast imaging employing steady-state acquisition or constructive interference into steady state, or T1-weighted 3-dimentional spoiled gradient recalled echo through the basal cisterns (to illustrate absence of connection between optic nerves) and ideally with high-resolution diffusion tensor imaging (to review the course and distribution of the optic tracts) are the preferred imaging protocols for diagnosis. The results of the ERG were normal. Although rare, achiasma should be considered in the differential diagnosis of congenital nystagmus. Thorough imaging evaluation is important to avoid misdiagnosis.

ARTICLE INFORMATION

Author Affiliations: School of Medicine, University of California, San Francisco (Sechrist); Department of Radiology & Biomedical Imaging, University of California, San Francisco (Glastonbury); Department of Ophthalmology, University of California, San Francisco (de Alba Campomanes).

Corresponding Author: Alejandra G. de Alba Campomanes, MD, Department of Ophthalmology, University of California, San Francisco, 490 Illinois St, San Francisco, CA 94158 (alejandra.dealba@ucsf.edu).

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REFERENCES

1. Hertle RW, Dell'Osso LF, FitzGibbon EJ, et al. Clinical, radiographic, and electrophysiologic findings in patients with achiasma or hypochiasma. *Neuroophthalmology*. 2001;26(1):43-57. doi:10.1076/noph.26.1.43.8055

2. EyeRounds. See-saw nystagmus: 64-year-old male who first noted the onset of oscillopsia five months prior to presentation. Accessed December 31, 2022. https://eyerounds.org/cases/case23.htm

3. Balani A, Kumar AD, Marda SS, Alwala S. Nondecussating retinal-fugal fiber syndrome: clinical and neuroimaging clues to diagnosis. *Indian J Ophthalmol*. 2015;63(11):858-861. doi:10.4103/0301-4738.171970

4. Bertsch M, Floyd M, Kehoe T, Pfeifer W, Drack AV. The clinical evaluation of infantile nystagmus: what to do first and why. *Ophthalmic Genet*. 2017;38(1):22-33. doi:10.1080/13816810.2016.1266667

5. Dumitrescu AV, Ashton Scruggs B, Drack AV. Clinical guidelines: childhood nystagmus workup. American Academy of Ophthalmology. Published February 12, 2020. Accessed February 17, 2023. https://www.aao.org/disease-review/clinical-guidelines-childhood-nystagmus-workup

6. Dell'Osso LF, Hertle RW, Williams RW, Jacobs JB. A new surgery for congenital nystagmus: effects of tenotomy on an achiasmatic canine and the role of extraocular proprioception. *J AAPOS*. 1999;3(3):166-182. doi:10.1016/S1091-8531(99)70063-7

7. Davies-Thompson J, Scheel M, Jane Lanyon L, Sinclair Barton JJ. Functional organisation of visual pathways in a patient with no optic chiasm. *Neuropsychologia*. 2013;51(7):1260-1272. doi:10.1016/j.neuropsychologia.2013.03.014

8. Prakash S, Dumoulin SO, Fischbein N, Wandell BA, Liao YJ. Congenital achiasma and see-saw nystagmus in VACTERL syndrome. *J Neuroophthalmol*. 2010;30(1):45-48. doi:10.1097/WNO.0b013e3181c28fc0

9. Pensiero S, Cecchini P, Michieletto P, Pelizzo G, Madonia M, Parentin F. Congenital aplasia of the optic chiasm and esophageal atresia: a case report. *J Med Case Rep*. 2011;5(1):335. doi:10.1186/1752-1947-5-335

10. Cochin JP, Hannequin D, Do Marcolino C, Didier T, Augustin P. Intermittent sea-saw nystagmus successfully treated with clonazepam. Article in French. *Rev Neurol (Paris)*. 1995;151(1):60-62.