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Two Additional Scenarios for See-saw Nystagmus: Achiasma and Hemichiasma

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The discovery of canine achiasma and hemichiasma has been followed by the identification of human achiasma (four individuals, to date). See-saw nystagmus was present in all cases of achiasma (canine and human) and in one of two cases of canine hemichiasma studied. Human infants with see-saw nystagmus should be imaged for possible structural abnormalities of the optic chiasm.
Key Words: Achiasma—Hemichiasma—See-saw nystagmus.

May and Truxal (1) have recently described see-saw nystagmus (SSN) in a patient with visual loss caused by cone–rod dystrophy. The patient had no abnormalities of the chiasm, midbrain, or thalamus. They mentioned two other recognized causes of SSN: congenital and mesodiencephalic lesions often associated with bitemporal hemianopia. The authors concluded by listing three scenarios for SSN: loss of vision, congenital disorder, or mesodiencephalic disease. We would add two additional scenarios: *achiasma* and *hemichiasma.*

Achiasma, first identified in a family of Belgian sheepdogs (2), is the congenital absence of an optic chiasm, totally precluding decussation of the retinal fibers from each eye and binocular vision. It does not result in bitemporal hemianopia, and no other midline abnormalities were present in the dogs or in the human diagnosed later. Achiasma is associated with congenital nystagmus and SSN. Hemichiasma is the uniocular failure of retinal-fiber decussation, despite the presence of an optic chiasm; it does not appear to preclude the development of binocular vision (3–6). One of us (L.F.D.) has been in the unique position to have studied congenital achiasma in dogs and in a human (7–9). Both the canine and human subjects had SSN, initially identified by video and direct examination, with waveforms later determined by eye-movement recordings (10–12).

After the above identifications, there have been reports of visual-evoked potential (VEP) data and preliminary eye-movement data from the human case (9,12–15). The VEP data suggest an abnormality in the number of crossing fibers opposite to that in albinism. Subsequent to the identification of canine achiasma, imaging of this human's optic chiasm resulted in the diagnosis of achiasma. Unfortunately, the preliminary eye movement data taken from the human patient were inadequate to identify SSN (13).

Most recently, postmortem anatomic studies showed that one of the canines with SSN had hemichiasma rather than achiasma (6). In a second anatomically documented case of canine hemichiasma, no nystagmus was present and the dog's behavior indicated good binocular function. Thus, one of two dogs with hemichiasma had both congenital nystagmus and SSN (mimicking achiasma), whereas the other had neither; both were capable of making uniocular saccades.

Based on the above studies, achiasma emerges as a sufficient condition for SSN, and SSN is a possible manifestation of hemichiasma. That is, all subjects with congenital achiasma, and some with hemichiasma, may have SSN. Although VEP asymmetry can suggest abnormalities in the number of crossing fibers (i.e., too many or too few), they cannot identify the structural abnormalities present in achiasma or hemichiasma; VEP asymmetry should alert the physician to the possibility of achiasma, which must then be verified by imaging. These new etiologies support the need for imaging studies of the optic chiasm in human infants with congenital SSN. Recently, Leitch et al. (16) diagnosed achiasma in a patient with midline craniofacial cleft and SSN, using MRI after recording abnormal VEP data. Imaging may also uncover septo-optic dysplasia, which is associated with SSN, although rarely (17,18). Unlike achiasma, it is also associated with bitemporal hemianopia, absence of the septum pellucidum, and other brain stem abnormalities.

An MRI should uncover the structural absence of a chiasm (achiasma) but may not be sufficiently sensitive

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to diagnose hemichiasma. With such imaging, we expect that additional incidences of achiasma will be diagnosed in humans. At present, there are no reported instances of human hemichiasma. However, because one of the two documented cases of canine hemichiasma did not manifest signs of visual dysfunction, cases of human hemichiasma may go undetected.

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