Diagnosis, assessment and management of nystagmus in childhood

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Abstract

Nystagmus is a rhythmical, repetitive and involuntary movement of the eyes. It is usually from side to side, but sometimes up and down or in a circular motion. It can be distressing to the child and their carers and leads to reduced visual acuity due to the excessive motion of images on the retina. It is helpful to distinguish between infantile nystagmus and acquired nystagmus. Infantile nystagmus usually appears by 3–6 months. Infantile nystagmus can occur with poor vision, or visual deprivation in early life due to congenital cataracts, optic nerve hypoplasia, retinal dystrophies or disease, can be associated to albinism or it can be part of a neurological syndromes or diseases. Neurological disease should be suspected when the nystagmus is asymmetrical or unilateral. Infantile nystagmus can be hereditary, the most common and best analysed form being a mutation of the *FRMD7* gene on chromosome Xq26.2. Novel methods to record eye movements and recent advances in the field of genetics have contributed to the improvement in diagnosis and management of nystagmus in children. Glasses, contact lenses and prisms help in improving vision. Pharmacological treatment exists to reduce nystagmus; however, as there are very few randomized controlled trials in this area, most pharmacological treatment options in nystagmus remain empirical. Eye muscle surgery may help in children with abnormal head posture to correct the defect and alleviate neck problems that can arise due to an abnormal head posture.

Keywords abnormal head posture; acquired nystagmus; albinism; infantile nystagmus; nystagmus; spasmus nutans

Introduction

Nystagmus is an involuntary rhythmic oscillation of the eyes. It can be caused by a slow drift of fixation which is followed by a fast refixation saccade (jerk nystagmus) or a slow movement back to fixation (pendular nystagmus). Nystagmus is a sign of a problem with the visual system or the pathways that connect the eyes to the parts of the brain that analyse vision. It can be easily identified by direct observation of eyes. Nystagmus is not painful and is not invariably associated with loss of vision.

Nystagmus is commonly encountered in clinical practice and the prevalence of nystagmus in general population is estimated to be 24 per 10,000 populations with a slight prediction to European ancestry. The prevalence of infantile nystagmus is 14 per 10,000. Nystagmus can be grouped as infantile nystagmus (IN), which usually appears in the first 3–6 months of life and acquired nystagmus (AN), which appears later.

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The child with nystagmus presents a diagnostic challenge. The concern is to ensure that the nystagmus is not a sign of significant neurologic abnormality which necessitates immediate intervention. The great majority of patients with nystagmus have an ophthalmic aetiology which can be elicited with simple examination techniques. There are certain types of nystagmus which do necessitate further intervention, and these are usually readily identifiable. While nystagmus can be confusing at first, a systematic approach beginning with a history and physical examination will often elicit its aetiology, and provide valuable information to the parents about the child's future development and visual prognosis.

Mechanism of nystagmus

To understand the mechanisms by which nystagmus may occur, it is important to recall the means by which the nervous system maintains position of the eyes. Foveal centration of an object of regard is necessary to obtain the highest level of visual acuity. Three mechanisms are involved in maintaining foveal centration of an object of interest: fixation, the vestibulo-ocular reflex, and the neural integrator. A failure of any part of this pathway can lead to nystagmus. However, the exact symptoms and signs will vary depending upon the underlying cause.

Fixation in the primary position involves the visual system's ability to detect drift of a foveating image and signal an appropriate corrective eye movement to refoveate the image of regard. Fixation requires reasonable visual acuity in the first place and this explains why impairment of vision can lead to nystagmus (see below).

The vestibulo-ocular reflex maintains foveation of an object during changes in head position. The proprioceptors of the vestibular system are the semicircular canals of the inner ear. Three semicircular canals are present on each side, anterior, posterior, and horizontal. The semicircular canals respond to changes in angular acceleration due to head rotation.

The neural integrator is a gaze-holding network of signals that aim to overcome the tendency of the eye to settle in the midline. When the eye is turned in an extreme position in the orbit, the fascia and ligaments that suspend the eye exert an elastic force to return toward the primary position. To overcome this force, a tonic contraction of the extraocular muscles is required. The cerebellum, ascending vestibular pathways, and oculomotor nuclei are important components of the neural integrator.

Causes of nystagmus in childhood

Nystagmus is classified as infantile or acquired nystagmus. Figure 1.

It is helpful to differentiate between infantile and acquired nystagmus. This can be done by considering the age of onset and waveform characteristics of nystagmus. The waveform can be formally obtained by eye movement recordings (techniques used include: electrooculography, scleral search coil and video eye-tracking devices).

The characteristics of the waveform include:

- Plane: horizontal, vertical, torsional, or any combination of these, such as seesaw nystagmus (vertical with torsional) or cyclorotatory nystagmus (horizontal with vertical).

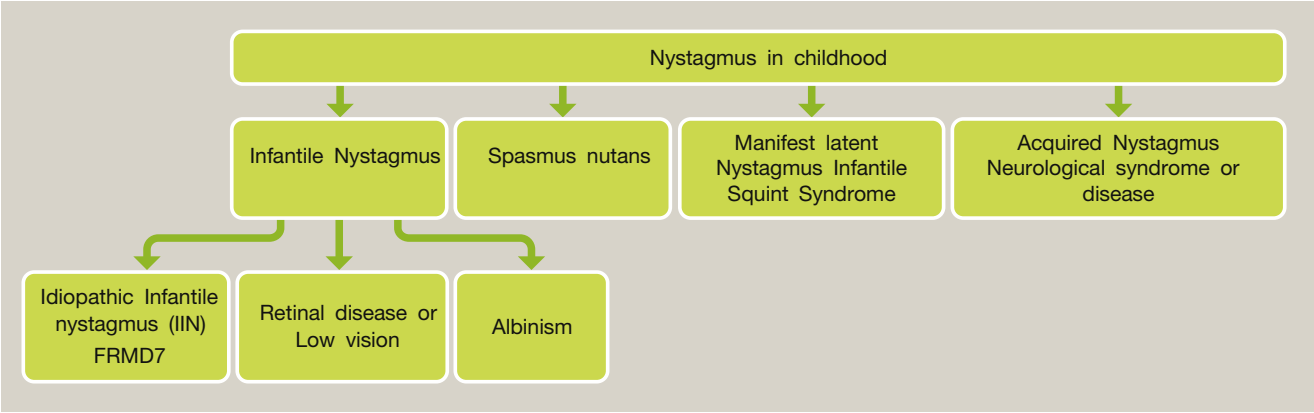

Figure 1 Classification of the different forms of childhood nystagmus.

- Amplitude (size) and frequency (cycles per second).
- Waveform: nystagmus has been divided into jerk nystagmus, which exhibits a quick and slow phase, and pendular nystagmus, which is a sinusoidal like oscillation without any obvious quick phase. In jerk nystagmus, the direction of nystagmus is defined by the quick phase of the jerk (e.g., downbeat). The slow phase can have accelerating and decelerating velocities (Figure 2).
- Conjugacy: when the eyes move in tandem, the nystagmus is described as conjugate or associated. Disconjugate or dissociated nystagmus occurs when the eye movements differ in amplitude, frequency, waveform, or when the oscillations of the two eyes are out of phase with each other e.g. Spasmus nutans, seesaw nystagmus, or convergence nystagmus in Parinaud syndrome.
- Foveation: many forms of congenital nystagmus show periods where the eyes move at a lower velocity allowing high-acuity vision at the fovea to function.
- Dependence on other parameters: certain types of nystagmus waveform are not constant but vary with time e.g. periodic alternating nystagmus is a horizontal jerk nystagmus that goes through cycles of left- and right-beating nystagmus, reversing approximately every 1–2 minutes. Eighty percent of nystagmus in infancy is infantile (congenital), and the remaining 20% is acquired.

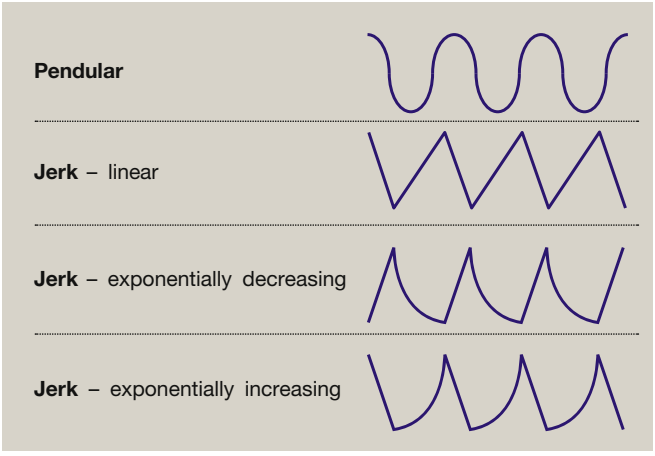

Figure 2 Waveform characteristics of different types of nystagmus.

Diseases affecting the visual system, such as retinal disorders causing visual loss, commonly lead to nystagmus because visual fixation is no longer possible. Disease affecting the vestibular organ in the inner ear causes an imbalance that leads to a mixed horizontal nystagmus, usually associated with vertigo. Disease affecting the central connections of the vestibular system, including the cerebellum, may cause several forms of nystagmus. These include down-beat, torsional, periodic alternating and see-saw nystagmus. None of these nystagmus types are, in themselves, pathognomonic of central nervous system disease. Nonetheless, down-beat nystagmus is usually associated with lesions of the vestibulo-cerebellum (flocculus, parafloccculus, nodulus and uvula) and the underlying medulla; up-beat nystagmus is most commonly reported with lesions of the medulla, including the perihypoglossal nuclei and adjacent vestibular nucleus (both structures are important for gaze-holding), the ventral tegmentum and the anterior vermis of the cerebellum; periodic alternating nystagmus is often linked to cerebellar disease (horizontal jerk nystagmus spontaneously reverses direction of the quick phase every few seconds); see-saw nystagmus is linked to parasellar lesions of the optic chiasm (e.g. pituitary tumours) and achiasma (rare form of pendular nystagmus in which the torsional components are conjugate and the vertical components are disjunctive (one eye rises and intorts while the other falls and extorts); and gaze evoked nystagmus is commonly seen as a side-effect of drugs, including sedatives, anticonvulsants and alcohol, as well as cerebellar disease.

Infantile nystagmus (IN)

Nystagmus associated with retinal diseases and low vision

Nystagmus associated to retinal diseases and low vision is more common than the infantile idiopathic nystagmus although both conditions have similar oculomotor features. Nystagmus in low vision is variable and it can be horizontal or vertical or a combination of both. It can be associated (conjugate) or dissociated.

The visual acuity is often lower than in idiopathic nystagmus due to the anatomical pathology of the eye. Nystagmus can result from congenital cataracts if not operated on early enough. Corneal opacities, developmental disorders of the optic disc and retina such as bilateral optic nerve hypoplasia and chorioretinal or optic nerve coloboma and retinopathy of prematurity are also associated with nystagmus. Deficits of rod and cone systems (congenital night blindness, achromatopsia), Leber's amaurosis,