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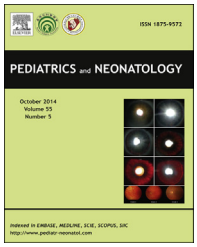

INVITED REVIEW ARTICLE

Nystagmus in Childhood

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Nystagmus is an involuntary rhythmic oscillation of the eyes, which leads to reduced visual acuity due to the excessive motion of images on the retina. Nystagmus can be grouped into infantile nystagmus (IN), which usually appears in the first 3–6 months of life, and acquired nystagmus (AN), which appears later. IN can be idiopathic or associated to albinism, retinal disease, low vision, or visual deprivation in early life, for example due to congenital cataracts, optic nerve hypoplasia, and retinal dystrophies, or it can be part of neurological syndromes and neurologic diseases. It is important to differentiate between infantile and acquired nystagmus. This can be achieved by considering not only the time of onset of the nystagmus, but also the waveform characteristics of the nystagmus. Neurological disease should be suspected when the nystagmus is asymmetrical or unilateral. Electrophysiology, laboratory tests, neurological, and imaging work-up may be necessary, in order to exclude any underlying ocular or systemic pathology in a child with nystagmus. Furthermore, the recent introduction of handheld spectral domain optical coherence tomography (HH SD-OCT) provides detailed assessment of foveal structure in several pediatric eye conditions associated with nystagmus and it can been used to determine the underlying cause of infantile nystagmus. Additionally, the development of novel methods to record eye movements can help to obtain more detailed information and assist the diagnosis. Recent advances in the field of genetics have identified the *FRMD7* gene as the major cause of hereditary X-linked nystagmus, which will possibly guide research towards gene therapy in the future. Treatment options for nystagmus involve pharmacological and surgical interventions. Clinically proven pharmacological treatments for nystagmus, such as gabapentin and memantine, are now beginning to emerge. In cases of obvious head posture, eye muscle surgery can be performed to shift the null zone of the nystagmus into the primary position, and also to alleviate neck problems that can arise due to an abnormal head posture.

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1. Introduction

Nystagmus is defined as an involuntary rhythmic oscillation of the eyes, and it can be confirmed relatively easily through direct observation of eyes and/or eye movement recordings. Nystagmus is commonly encountered in clinical practice and leads to reduced visual acuity due to the excessive motion of images on the retina, and also the movement of images away from the fovea.[1] The prevalence of nystagmus in the general population is estimated to be 24 per 10,000 population with a slight predilection toward European ancestry.[2] The prevalence of infantile nystagmus is 14 per 10,000.[2]

Nystagmus can be grouped into infantile nystagmus (IN), which usually appears in the first 3–6 months of life, and acquired nystagmus (AN), which appears later. For those who have infantile nystagmus (IN), this can be idiopathic or associated to another eye disease, such as retinal disease, albinism, low vision, or visual deprivation in early life (due, for example, to congenital cataracts or optic nerve hypoplasia). Nystagmus can also be part of neurological syndromes and neurologic diseases (Figure 1).[3] In IN associated with other eye diseases, vision is not only affected by the excessive motion of the image on the retina caused by the nystagmus, but also by a defective visual system.[4] Mechanisms underlying IN are not very clear. Numerous hypotheses and models have been proposed to explain the ocular oscillations observed in IN, usually highlighting various elements of the ocular motor circuitry as the direct cause.[5] However, the clear association between IN and the many sensory anomalies that lead to sight loss during visual development imply an afferent cause to many IN forms.[6]

Acquired nystagmus can result from a range of neurological disorders, of which the most common are multiple sclerosis, disease of the vestibular apparatus and innervations, insult to the nervous system caused by stroke, tumors, or trauma, and as a result of drug toxicity.[7] In general, the mechanisms underlying AN are better understood than those behind IN. AN is commonly associated with lesions to the subcortical ocular motor circuitry.[8] Consequently, detection and diagnosis of diseases associated with AN are greatly assisted by neuroimaging methods.

Nystagmus can be distressing for both those with IN and AN. Nystagmus leads to deterioration in visual acuity and motion sensitivity mainly because of deterioration in foveal vision, when images move across the retina rapidly.[9] Nystagmus can also have a significant psychological and social impact.[10] Although patients with IN and AN both have reduced vision, patients with AN tend to suffer from oscillopsia (the illusion of constant movement of the surroundings) and hence may be more troubled by the condition.[11] Patients with IN do not usually suffer from oscillopsia. Two hypotheses have been suggested to explain the mechanism behind the suppression of oscillopsia. The first is the "sampling theory", by which the information from the most stable retinal images during the foveation periods can be used to establish a stable image, whereas the rest of the nystagmus cycle is ignored.[12] The second hypothesis is the "remapping theory" whereby an efference copy signal of the nystagmus waveform is used to cancel the effects of motion.[13] This is probably the most likely theory because vision during the fast phases of the nystagmus cycle has been documented which argues against sampling. Finally, a recent Positron Emission Tomography/Magnetic Resonance Imaging (PET/MRI) study in a patient with infantile nystagmus during maximal nystagmus showed a metabolic downregulation of the area MT/V5 bilaterally, the most important part of the visual cortex for visual motion

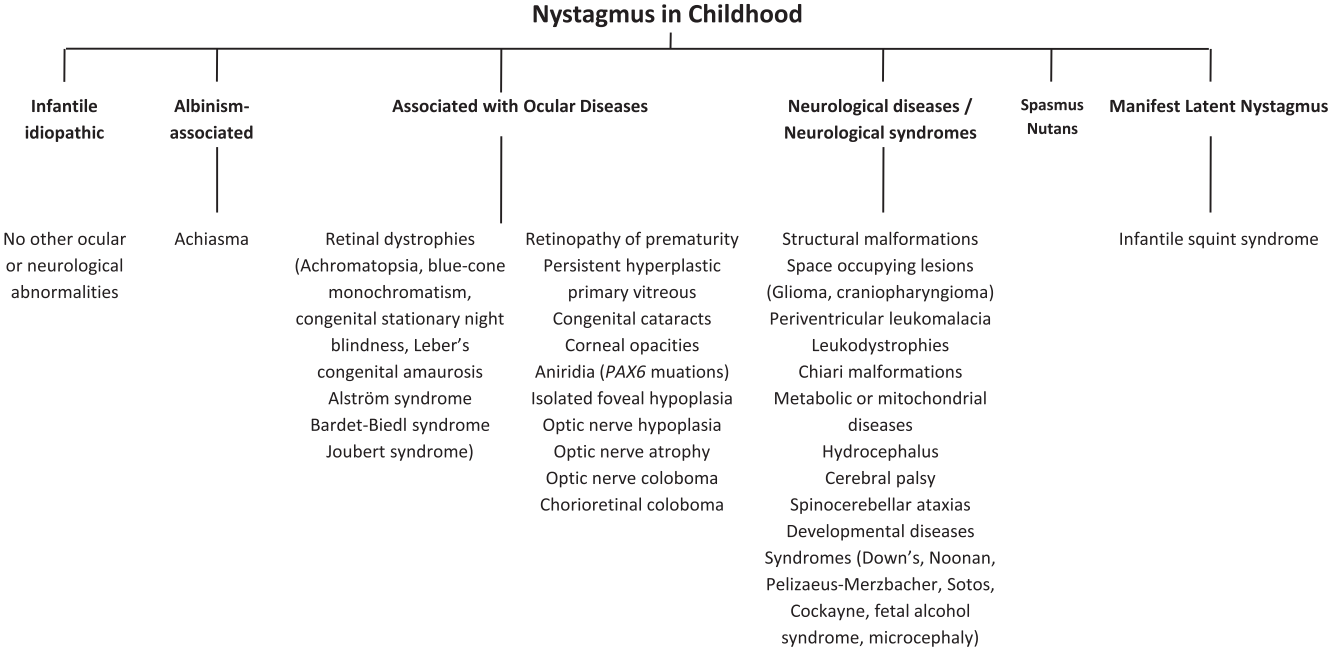

Figure 1 Classification of childhood nystagmus. *Note*. From "The pharmacological treatment of nystagmus: a review", by R.J. McLean and I. Gottlob, 2009, *Expert Opin Pharmacother*, *10*, p. 1805–16. Copyright 2009, *Informa UK Limited*. Adapted with permission.