

Congenital nystagmus

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[Abstract](#)
[Key-words](#)
[Disease name](#)
[Definition/Diagnosis criteria](#)
[Differential diagnosis](#)
[Pathophysiology](#)
[Clinical description](#)
[Paraclinic testing and diagnosis](#)
[Frequency](#)
[Treatment](#)
[Reference](#)

Abstract

Idiopathic congenital nystagmus is defined as conjugated, spontaneous and involuntary ocular oscillations that appear at birth or during the first three months of life. This nystagmus persists throughout life. The frequency is estimated to 1 in 1500 births. Ocular oscillations are often symmetric and usually horizontal in 95% of patients. They can persist after eye closure, however, decrease of idiopathic congenital nystagmus have been reported during non-visual tasks. Nystagmus can be classified into different categories (Pendular nystagmus, horizontal unidirectional nystagmus, bi-directional nystagmus) according to the characteristics of their oscillations: peak-to-peak amplitude, frequency, mean velocity, direction and period of foveation. This disorder is believed to be due to a primary abnormality in oculomotor control. Autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive modes of inheritance have been described. The gene that maps to chromosomes 6p12 (NYS2) is associated with autosomal dominant inheritance. The genes mapped to chromosome Xp11.4-p11.3 (NYS1) and Xq26-q27 are associated with X-linked congenital forms. There are evidences for a fourth gene in idiopathic congenital nystagmus. Complete ophthalmologic examination and electrophysiological study must be performed to rule out any ocular abnormalities causing bilateral visual deprivation or retinal dysfunction. Treatment of idiopathic congenital nystagmus aims at improving the vision. It includes correction of refractive errors, drugs and eye muscle surgery.

Key-words

Idiopathic congenital nystagmus, foveation periods, *NYS2*, *NYS1*, Xq26-q27

Disease name

Congenital motor nystagmus
 Idiopathic congenital nystagmus

ocular oscillations that appear at birth or during the first three months of life. This nystagmus persists throughout life.

Definition/Diagnosis criteria

Idiopathic congenital nystagmus is defined as conjugated, spontaneous and involuntary

Differential diagnosis

Idiopathic congenital nystagmus must be distinguished from sensory congenital

nystagmus associated with congenital retinal disease due to genetic disorder or with other abnormal conditions causing deprivation of the visual system. The most frequent excluded diseases include aniridia with macular hypoplasia, all forms of albinism, early retinal dystrophies, congenital stationary night blindness which maps to chromosome Xp11.3, achromatopsia, blue cone monochromatism which maps to chromosome Xq28, bilateral optic nerve hypoplasia, chorio-retinal or papillary coloboma, recessive optic atrophy. Ophthalmologic examination and electrophysiological studies can easily rule out these retinal and optic nerve pathologies. In addition, sensory congenital nystagmus may result from bilateral congenital cataracts, which are also easily excluded with ocular examination. Central nervous system dysfunction must also be ruled out, since neurological abnormalities are not encountered in idiopathic congenital nystagmus.

Pathophysiology

Different hypotheses have been proposed to explain the mechanisms involved in idiopathic congenital nystagmus. A defect in motor control of visual fixation or an abnormal development of fixation system of the brain without any detectable central nervous system abnormalities, as well as an instability of the neural integrator responsible for gaze holder may underlie the pathology. However, fixation mechanisms appear to be functional in congenital nystagmus since it is associated with strong fixation reflexes. In addition, idiopathic congenital nystagmus can increase during fixation and decrease in non-visual tasks. Thus, it has been suggested that this nystagmus can result from an abnormal circuit between the fixation system and ocular stabilization systems. However, it is not yet possible to link the waveform presentation in idiopathic congenital nystagmus with an etiology or an ocular motor system dysfunction.

Autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive modes of inheritance have been described. However, the existence of dominant and recessive forms in X-linked inheritance has not been clearly established. In addition, penetrance could be incomplete in these X-linked forms.

At least, 4 genes are suspected to be associated with idiopathic congenital nystagmus. The gene that maps to chromosome 6p12 (*NYS2*) is associated with autosomal dominant inheritance. The genes mapped to chromosome Xp11.4-p11.3 (*NYS1*) and Xq26-q27 are associated with X-linked

congenital nystagmus. However, in some families, with male-to-male transmission, linkage to chromosome 6 was excluded, providing evidence for a fourth causative gene. There is no candidate gene associated with these different loci.

Clinical description

Idiopathic congenital nystagmus is characterized by conjugated, bilateral, spontaneous, involuntary and uncontrollable ocular oscillations. These oscillations are often symmetric and usually horizontal in 95% of patients. Oscillations can persist after eye closure. However, decrease of idiopathic congenital nystagmus have been reported during non-visual tasks. Nystagmus can be classified into different categories according to the characteristics of their oscillations: peak-to-peak amplitude, frequency, mean velocity, direction and period of foveation.

Pendular nystagmus is a rare form of idiopathic congenital nystagmus and is more specific of youngest patients. Most of the patients, especially after the age of 18 months, present with a jerk nystagmus, with accelerating slow phase in eccentric gaze.

Horizontal unidirectional nystagmus is rare in idiopathic congenital nystagmus and characterized by oscillations beating always in the same direction. A *bi-directional nystagmus* is most often encountered, as the direction of oscillation changes according to gaze direction. Nystagmus beats to the right when gaze is oriented to the right and to the left when gaze is oriented to the left. In reversing zone of nystagmus direction, oscillations slow down with reduced amplitude or disappear. Thus, visual acuity is better in this null zone and patients try to use it, inducing a torticollis. Null zone can be localized in primary position, in an eccentric position of gaze or in convergence. Some patients present with two null zone, one in each gaze direction.

Visual acuity and contrast sensitivity are reduced during nystagmus, due to the continuous shifting of images on the fovea that is induced by ocular oscillations. In contrast to what is observed during primary sensory nystagmus, visual acuity is usually better preserved in patients with idiopathic congenital nystagmus. and is sometimes normal. Visual acuity ranges from 20/50 to 20/20, with a mean value of 20/30. However, visual acuity is related to the duration of foveation periods. Foveation periods are well preserved during idiopathic congenital nystagmus since slow phases velocity allows the fixation system to prolong the fixation and the foveation when the images fall on the fovea. The longer the

foveation time the higher the visual acuity. In addition, a relationship exists between the visual acuity and the eye position during foveation. A variability of eye position is responsible for a decrease of visual acuity in spite of long foveation periods. Patients do not perceived oscillopsia in idiopathic congenital nystagmus except when new visual sensory conditions appear such as strabismus.

Paraclinic testing and diagnosis

Complete ophthalmologic examination must be performed to rule out any ocular abnormalities causing bilateral visual deprivation or retinal dysfunction. However, even when ocular examination is normal, an electrophysiological study with electroretinography and visual evoked potentials is always necessary to affirm the absence of retinal or optic tract dysfunction. Neuro-imagery can be required in case of nystagmus appearing during months after birth and without any evidence of ocular pathology or electrophysiological abnormality. Eye movement recordings are useful to characterize the nystagmus. They provide information on amplitude, frequency and mean velocity as well as the aspect or direction of the waveforms. Triangular waveforms are most often encountered in younger children and pendular nystagmus in patients less than 18 months.

Frequency

The frequency of idiopathic congenital nystagmus is estimated to 1 in 1500 births. X-linked mode of inheritance is probably the most common form of idiopathic congenital nystagmus.

Treatment

The treatment of idiopathic congenital nystagmus aims at improving the vision. All refractive errors must be first corrected, and require an objective refraction after cycloplegia. Optic lens are often a better way to correct these refractive errors since they move with the eyes during ocular oscillations. However, prismatic spectacles can be used when the null region is eccentric, in order to shift the visual scene laterally and to reduce the torticollis. But, their indication is limited to small eccentrication of the null position due to reduction and distortion of vision they induce when their value is too important. The base of the prisms must be in the same direction on both eyes, away from the null position. Base out prisms may be used to damp idiopathic congenital nystagmus during distance fixation in patients with null zone in convergence.

Other non-surgical techniques have been proposed, such as the afferent stimulations of the ophthalmic division of the trigeminal nerve or cutaneous stimulations. Pharmacological treatment of idiopathic congenital nystagmus has limited interest in the literature. An inhibitor of glutamate release, the Baclofen, can improve the visual acuity in small series of patients.

In cases of high eccentrication of the null position, surgery for head posture can be achieved. Kestenbaum proposed to perform recessions and/or resections of the four horizontal muscles in order to shift horizontally the null position in primary position. Recessions and/or resections of the four vertical muscles have been proposed when the null zone must be shifted vertically. In torsional null zone, the Spielmann procedure consists in a slanting or the insertion of the rectus muscles.

In order to improve visual acuity, different authors have proposed to perform a large recession of all horizontal muscles. Such procedures decrease the intensity of the oscillations of idiopathic congenital nystagmus. A good result however is difficult to obtain and iterative surgical procedures are often necessary.

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