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Reviving the differential diagnosis of congenital nystagmus

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Background: Irreversible nystagmus of infancy is one of the main diagnostic clinical signs in some important neurologic syndromes. It could help physicians to locate the site of injury.

Purpose of the article: In this article we hope to add a new path for differential diagnosis of congenital nystagmus.

Main finding: In the available medical literature primary nystagmus was found to be a main component of the Charcot's triad for the diagnosis of immune-mediated Multiple Sclerosis; composed of nystagmus, Intention tremor and staccato speech, and of the recently published "Mais-Nadim Nasser triad", dedicated for Leukodystrophies (LDs) diagnosis, which is composed from: Primary hypotonia, irreversible nystagmus and abnormal BERA test, as well. When nystagmus manifests in an infant, along with the emergence of the signs mentioned above: All or some of them, it is a warning signal for serious diseases, and help early diagnosis of white matter disorders. These triads should be a critical part of the differential diagnosis of congenital nystagmus. Other metabolic and genetic syndromes have a very low incidence of nystagmus.

Conclusions: Nystagmus in early life should alert the ophthalmologist for existing serious diseases of the central nervous system, particularly, hypo or demyelination of brain white matter; the more so when it is manifests along with hypotonia and deafness, or by intention tremor and staccato speech. We advise that these features, together with nystagmus, be inserted as a new track into the algorithm of differential diagnosis of nystagmus.

Summary: Irreversible Infantile nystagmus is a very suggestive sign of white matter disease in the central nervous system, especially if it is accompanied with hypotonia and abnormal BERA test, tremor or interrupted speech, and we intend to set them as an essential element of nystagmus diagnostic algorithms.

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