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Perspective

Neurometabolic Diseases with Eye Movement Disorders in Child

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ABOUT THE STUDY

Neurometabolic diseases are sporadic genetic disorders caused by reduced enzyme activity and a lack of cofactors or transporters, which can affect protein, carbohydrate, and lipid metabolisms or directly affect energy supply. There have been over 750 neurometabolic disorders documented. Each illness has a low prevalence, but when combined, the cumulative frequency is as high as 1 in 784. The majority (72%) are diagnosed by the age of 15, and one-third by the age of one year. The cerebral cortex, basal ganglia, brainstem, cerebellum, vestibular system, and motor units for cranial nerves and muscles are all involved in the eye movement network, as well as all the interconnections between them. The frontal eye field, supplemental eye field, dorsolateral prefrontal cortex, and parietal cortex are all involved in voluntary regulation of eye movements.

During prenatal through adulthood, neurometabolic disorders are generally defined by a deficiency of particular substrates or an accumulation of toxins or metabolites. Neurometabolic illnesses can be divided into small molecule disorders, big molecule disorders, disorders of brain energy metabolism, and miscellaneous diseases on a pathophysiologic level. The majority of the clinical characteristics are progressive neurodegenerative illnesses. Impaired mitochondrial function or glucose transporter abnormalities are examples of cerebral energy metabolism diseases. Clinical symptoms are frequently accompanied by mobility abnormalities and manifest as chronic worsening or episodic aggravation. Disorders of metabolism involving carbohydrates, purines, pyrimidine's, heavy metals, and biogenic amine neurotransmitters are among the other miscellaneous conditions.

Small molecule disorders

Maple Syrup Urine Disease (MSUD): In the first week of birth, patients with typical MSUD exhibit poor feeding, lethargy, and opisthotonus. Dystonia, ataxia, and choreoathetosis are all possible symptoms in children and toddlers. Ophthalmoplegia in the newborn phase has been recorded, as well as bilateral adduction paresis and lack of upward vision with slight ptosis. Optic atrophy, nystagmus, ophthalmoplegia, and cortical blindness have all been observed in untreated individuals. Diffuse cerebral edoema including the basal ganglia, cerebellum, mesencephalon, pons and supratentorial, and thalamus is a common finding on Magnetic Resonance Imaging (MRI) in newborn MSUD.

Nonketotic hyperglycemia: In neonatal periods, the typical neonatal form of NKH is characterized by lethargy, coma, myoclonic seizure, hiccup, and apnea. Intellectual impairment, axial hypotonia, and spasticity are other common side effects. Patients with late-onset atypical NKH (symptoms that appear after the age of two) have a more diverse and milder phenotype.

Large molecule disorders

Gaucher disease: Gaucher syndrome causes glucosylsphingosine to accumulate in the viscera and brain, particularly the brainstem, deep cerebellar nuclei, thalamus, and basal ganglia. The ocular motor expression is only seen in neuropathic gaucher illness and is one of the first symptoms. Eye movement issues appeared by the age of two in one cross-section study of neuronopathic gaucher illness. Oculomotor apraxia with delayed start, sluggish saccade, and horizontal supranuclear gaze palsy are the most prevalent eye motor symptoms. Vertical saccade dysfunction may manifest later, with downward saccades being affected more than upward gazing.

Niemann-Pick disease type C (NPC): Hepatosplenomegaly, cholestasis, developmental delay, and progressive dementia are some of the visceral, neurodegenerative, and psychological symptoms that patients may experience. Symptoms appear in the majority of individuals throughout their youth. In individuals with neurological symptoms, cerebellar ataxia, dysmetria, dysarthria, dysphagia, and gelastic cataplexy are common neurological indicators. Vertical gaze palsy affects individuals of practically every age group. Although brain MRI abnormalities are non-specific, aberrant results have been observed in the cerebellum, hippocampus, and subcortical grey matter volume decreases, as well as mild changes in the white matter areas.

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Zellweger spectrum disorders: Craniofacial dysmorphism, cataracts, retinopathy, hepatic illness, renal cysts, adrenal hypoplasia, neural migration disorder, and leukodystrophy are all symptoms of that kind of disorder. In the majority of instances, death occurs during the first year of life. The clinical history of newborn adrenoleukodystrophy and infantile refsum illness is similar but milder. They may live into childhood or, in exceptional cases, adulthood. In individuals with extensive demyelination of the subcortical regions and brainstem, pendular nystagmus is a common eye motor abnormality in Zellweger spectrum illnesses. Patients with gaze-evoked nystagmus and square-wave jerks with saccadic intrusions may have a moderate phenotype of Zellweger spectrum diseases with substantial cerebellar atrophy.

Congenital Disorders of Glycosylation (CDG): Patients with CDG-Ia have hypotonia, esotropia, inverted nipples, aberrant fat distribution in the buttocks and suprapubic regions, and intellectual impairment. Endocrine and coagulation problems are possible in patients. In most individuals, the cerebellum is damaged, manifesting as cerebellar hypoplasia at birth or subsequently as progressive cerebellar atrophy. Ataxia, dysmetria, and truncal titubation are all possible symptoms. Ocular flutter, square-wave jerks superimposed over a horizontal pendular nystagmus, and horizontal ocular motor apraxia are examples of eye motor disorders. Smooth pursuit and optokinetic response in both vertical and horizontal planes may be hindered in patients.

Cerebral energy metabolism disorders: Patients present with a wide phenotypic spectrum of symptoms, including epilepsy, mobility abnormalities, and cognitive impairment. Atypical eye movements, such as paroxysmal eye–head motions, might be a symptom of glucose transporter type 1 impairment. The event usually appears around the age of 6 months and disappears by the age of 8, with each incident lasting about 5 minutes. Head movement in the same direction as eye movement is referred to as eye–head movement.

Mitochondrial diseases: Cataract, pigmentary retinitis, optic atrophy, ptosis, and ophthalmoplegia are all common ophthalmic symptoms of mitochondrial disorders. The most prevalent ocular motor manifestation of mitochondrial disorders is chronic progressive ophthalmoplegia, which is characterized by progressively progressing bilateral ocular immobility. Because of the sluggish development and near-absence of diplopia, the majority of cases went unreported. Pyruvate dehydrogenase is a multi-enzyme complex found in mitochondria that catalyzes the conversion of pyruvate to acetyl coenzyme A. Young infants with pyruvate dehydrogenase E2 impairment have been observed to develop nystagmus or eye rolling. Eye movement problems may be seen in other mitochondrial illnesses impacting the brainstem or cerebellum.

Short chain acyl coenzyme A deficiency: Internal ophthalmoplegia, developmental delay, microcephaly, and myopathy, as well as external ophthalmoplegia, are all clinical indications of this condition, which is caused by defective fatty acid beta-oxidation.