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Cerebellar Ataxia Infantile with Progressive External Ophthalmoplegia

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Perspective

A rare genetic, neuro-ophthalmological disease characterized by progressive weakness of the external eye muscles, resulting in bilateral ptosis and diffuse, symmetric ophthalmoparesis. Additional signs may include generalized skeletal muscle weakness, muscle atrophy, sensory axonal neuropathy, ataxia, cardiomyopathy, and psychiatric symptoms. It is usually more severe than autosomal dominant form. Making a diagnosis for a genetic or rare disease can often be challenging. Healthcare professionals typically look at a person's medical history, symptoms, physical exam, and laboratory test results in order to make a diagnosis. The following resources provide information relating to diagnosis and testing for this condition. If you have questions about getting a diagnosis, you should contact a healthcare professional. Chronic progressive external ophthalmoplegia is a slowly progressive extraocular muscle disorder characterized by bilateral, usually symmetrical, limitation of eye movements, and ptosis.

This classical manifestation of mitochondrial disease can develop either in isolation or, more frequently, in association with other multisystemic features. An expanding list of nuclear-encoded mitochondrial genes is now known to cause CPEO phenotypes, predominantly in the context of multiple mitochondrial DNA deletions, but also as part of mtDNA depletion syndromes. This genetic heterogeneity can result in diagnostic delays, which is further compounded by the clinical challenges inherent in distinguishing CPEO from other disorders that also present with external ophthalmoplegia and ptosis.

Corrective ptosis surgery can be a highly effective treatment and alleviating symptoms of diplopia can make a significant difference to the individual's quality of life. Ophthalmoparesis develops over many years and may lead to complete ocular paralysis. Additionally, many patients have some weakness of the orbicularis muscle. Some patients seek medical attention only when ptosis covers the optic axis and leads to visual disturbance. Patients with CPEO use their frontalis muscles to lift their eyelids and show compensatory chin elevation. Together with the ptosis, this is called Hutchinson triad. Ophthalmoplegia is often symmetric and may not lead to complaints because patients simply turn their heads to compensate. Only a minority of patients suffer from diplopia. Muscle weakness is often not restricted to the extraocular muscles, and severe weakness of the facial muscles can present with the facies myopathica. Many patients also suffer from exercise intolerance.

In most cases, neurologic examination shows limb weakness, most prominent in the proximal muscles of the lower extremities. Additionally, involvement of systems other than muscle is possible in CPEO. The term CPEO-plus was established for these patients. Ocular manifestations include retinopathy, optic atrophy, and, rarely, cataracts. Cardiac manifestations include cardiac conduction block and cardiomyopathy. Cerebral manifestations include epilepsy, cerebellar ataxia, and dementia. The peripheral nervous system can also be affected, typically with axonal sensory neuropathy. Endocrine involvement includes diabetes mellitus, hypothyroidism, hypoparathyroidism, and hypogonadism. Sensorineural hearing loss and gastrointestinal involvement are also possible. Chronic PEO is an adult-onset disease characterized by bilateral ptosis, ophthalmoplegia, and proximal myopathy in which mild cerebellar ataxia can also occur.

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