

Eyelid drooping may exist in rare diseases of muscle and neuromuscular junction

Eyelid drooping or ptosis is due to many different possible causes, ranging from natural causes (eg natural aging process) to more serious conditions. The ability to open the eye depends on a muscle in the orbit, named levator palpebrae superioris (latin of elevating muscle of upper eyelid), that elevates and retracts the upper eyelid.

In this literature review, we focus on ptosis (isolated or associated with other symptoms) in rare neuromuscular diseases, with the aim to propose a diagnostic approach when a rare myopathy is suspected.

There is no universal definition of a rare disease. However, medical authorities around the world generally base on a low prevalence for assigning the status of rare disease. The upper limit accepted in Europe is of one person out of 2,000. Rare neuromuscular diseases refers to a heterogeneous group of genetic or acquired diseases. Acquired diseases may be of autoimmune origin. Ptosis in rare neuromuscular disorders warrant close collaboration between ophthalmologists and neurologists. When the diagnosis of a rare myopathy is known, eye examination is essential, as treatments to limit visual impairment may be proposed. Otherwise, eyelid drooping, sometimes discrete, needs to be recognized by the ophthalmologist as a marker of myopathy. Damage of the levator muscle of upper eyelid must be distinguished from damage of neuromuscular transmission in neuromuscular junctions.



Fatigue (Ptosis) in a patient with Myasthenia Gravis (in an article of Posey & Spiller, ed.1904).
(Original text : Posey & Spiller, public domain, authors are both more than 70 years death.)

Myasthenia gravis (MG) is an example of neuromuscular junction disorder, characterized by weakness and fatigability of muscles. MG is an autoimmune disease where anomalous antibodies

are produced against the acetylcholine receptors in voluntary muscles. MG may be limited to the muscles of the eye (ocular MG), leading to abrupt onset of weakness/fatigability of the eyelids or eye movement. MG may also involve other skeletal muscle groups (generalized MG). A drooping eyelid can stay constant, worsen over time (be progressive), or come and go (be intermittent).

Ptosis is fluctuating and asymmetrical (if bilateral) in rare diseases of neuromuscular junction, It occurs with or without diplopia, and may be indicative of myasthenia gravis in 40-50% of cases; whereas in congenital myasthenic syndromes, ptosis is rarely a prevailing sign but its presence is a valuable clinical clue. In Lambert-Eaton myasthenic syndrome, ptosis is rather moderate and bilateral. In rare myopathies (with involvement of the levator muscle of upper eyelid), ptosis is usually an isolated ocular sign, and bilateral in congenital myopathies, glycogen storage disease type II (also called Pompe disease or acid maltase deficiency). A unilateral ptosis associated with retroorbital pain and peri-orbital edema evokes myositis.

In ocular pharyngolaryngeal muscular dystrophy, ptosis is the most common initial symptom, followed by swallowing difficulties (dysphagia) and muscle weakness. Dysphagia begins with food but as the condition worsens liquids become difficult to swallow as well. Limb weakness develops later on in the disease. Ptosis associated with ophthalmoplegia (paralysis or weakness of one or more of the six muscles that hold the eye in place and control its movement) may suggest a mitochondrial myopathy.

In myotonic dystrophy, ptosis is frequent and asymmetric, even if it is not marked and causing little discomfort to the patient.

The diagnostic tests, such as dosage of CPK (creatine phosphokinase), EMG (electromyogram), muscle biopsy and genetic studies, are used to confirm the diagnosis.

A rational diagnostic approach is needed to avoid unnecessary examinations. It will implement a therapy and/or a suitable support without delay, and if necessary, adequate genetic counseling.

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[\[Ptosis in rare muscle and neuromuscular junction disorders: A literature review and diagnostic flowchart\].](#)

[\[Article in French\]](#)

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