Diplopia demystified: a global diagnostic approach from brain to muscle
LOOK AT THE EYES: diplopia and neuromuscolar disorders

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No disclosures

Extrinsic ocular muscles have specific structural, functional, biochemical-metabolic, and immunological characteristics distinct from the other skeletal muscles

- ❖multiply innervated
- ❖fast
- ❖rich in mitochondria (high oxidative capacity)

they

- co-express multiple myosin heavy chain isoforms in addition to embryonic myosin
- have very high fatigue resistance
- have ability to quickly and precisely control ocular motility

The extraocular muscles, due to their unique characteristics, are selectively spared in aging and in some forms of muscular dystrophy (e.g. Duchenne or some forms of LGMDs…) -> myogenic cell precursor populations differ from those of the limbs, with a higher proliferative capacity and continuous remodeling,

and vice versa

they are typically and commonly affected in specific diseases:

- Myasthenia Gravis
- Mitochondrial Myopathies
- Ophthalmopathies associated with thyroid dysfunction
- Overlap syndromes between the above (differential diagnosis sometimes difficult)

Differential Diagnosis

Myogenic

- mitochondrial diseases
- oculopharyngeal muscular dystrophy
- congenital myopathies
- thyroid orbitopathies
- ocular myositis

Neurogenic

- congenital fibrosis of the oculomotor muscles
- Miller-Fisher syndrome
- diabetic neuropathy
- other rare motor neuron/peripheral nerve conditions

Neuromuscular juntion diseases

- autoimmune Myasthenias
- congenital Myasthenias
- Botulism

Diagnosis

Hereditary diseases often lead to ophthalmoparesis without diplopia, while in acquired diseases ophthalmoparesis is often asymmetrical. Diplopia and fluctuation are the hallmark of an acquired synaptic disease.

Diagnostic tests

- EMG, neurography
- SRS, single-fiber EMG
- thyroid hormones, CK
- specific antibodies
- lactic acid
- muscle biopsy
- brain and muscle MRI

Learning objectives

- recognize diseases, making thoughtful differential diagnosis
- acquire basic elements to threat patients

Supported by presentation of videos and discussion of clinical cases

Take home message

- LOOK AT THE EYES: it could provide valuable clues in the differential diagnosis of neuromuscolar disorders

➢ correct diagnosis means correct therapy: there are many rules, but also many exceptions

➢ Don't miss treatable disorders (symmetric opthalmoplegia is not always a mitochondrial myopathy…)

➢ Don't overthreat untreatable diseases (asymmentric ocular involment and dyplopia are not always due to myasthenia gravis)