

Neuromuscular Disorders (GENERAL)

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NEUROMUSCULAR DISEASES - disorders of **motor unit** (specifically excluded are suprasegmental disorders!):

- 1) lower motor neuron / its axon – **neurogenic disorders**
- 2) neuromuscular junction - **neuromuscular junction disorders**
- 3) muscle fibers – **myogenic disorders**

CLINICAL MANIFESTATIONS

1. Symmetrical flaccid muscle **weakness**
2. Muscle **wasting / atrophy** or (**pseudo**)**hypertrophy**
Advisable to use term “wasting” in **myogenic disorders** and “atrophy” in **neurogenic disorders!**
3. **Hypo(a)reflexia**
4. **Fasciculations** (specific for **neurogenic disorders**) see p. Mov3 >>
5. **Sensory changes** (specific for **neurogenic disorders**)
6. **Myotonia** (specific for **myogenic disorders**); may be accompanied by muscle hypertrophy.
7. Painful **cramps** (esp. in **LMN disorders**; not in **neuromuscular junction disorders**).
see p. Mov3 >>
8. **Myalgias**
9. **Muscle contractures** (**glycolytic enzyme defects**) - last longer than cramps, provoked by exercise, electrically silent. see p. Mov3 >>
10. Tendon **contractures** (in **myopathies** of long duration; early contractures - Emery-Dreifuss dystrophy, Bethlem myopathy).
11. Bladder disorders (in **neurogenic disorders**)

NEUROGENIC disorders:

- 1) initially affects **DISTAL muscle groups** (with exceptions – see below).
- 2) **atrophy** > weakness
- 3) **reflexes early absent**
- 4) **fasciculations** ± sensory changes.
- 5) possible **myalgias** (e.g. Guillain-Barré syndrome), **painful cramps**
- 6) possible **contractures of muscles**
- 7) possible **bladder disorders**

NEUROMUSCULAR JUNCTIONAL disorders:

- 1) **variable fatigable weakness** (initially in **EXTRAOCULAR & BULBAR muscles**)
- 2) **no atrophy!!!**
- 3) **reflexes present!!!**
- 4) no fasciculations, no sensory loss.

MYOGENIC disorders:

- 1) initially affects large **PROXIMAL muscle groups** (with exceptions – see below); neck flexion is much weaker than neck extension.
- 2) **weakness** > atrophy
- 3) **reflexes long present** (diminished in proportion to weakness degree)
- 4) no fasciculations, no sensory loss.
- 5) possible **myalgias** (surprisingly uncommon in most muscle diseases!), **painful cramps**
- 6) possible **myotonia**
- 7) possible **contractures of muscles**
- 8) possible **myoglobinuria**

CONGENITAL neuromuscular disease:

- decreased **fetal movements**.
- intrauterine **growth retardation, low weight** for gestational age (because of *small muscle mass*).
- generalized **hypotonia**.
- funnel shape **thorax**, thin radiolucent **ribs** (due to *intercostal muscle weakness*).
- male infants may have **undescended testicles** (due to weak *gubernaculum*).
- **developmental delay**.

Try to establish PATTERN OF WEAKNESS:

1. **LIMB-GIRDLE weakness** (most common and therefore least specific pattern) - weakness exclusively / predominantly in **limb proximal muscles**; neck flexor & extensor muscles can also be affected; etiology – *many myopathies*, some *neuropathies*:
 - *Kugelberg-Welander syndrome* – affects proximal limbs
 - *acquired demyelinating neuropathies* (Guillain-Barré syndrome, chronic inflammatory demyelinating polyneuropathy) may have proximal as well as distal involvement!
2. **DISTAL EXTREMITY weakness** in upper extremities (extensor muscle group) or lower extremities (anterior or posterior compartment muscle groups); etiology:
 - 1) *neuropathies* (most commonly)
 - 2) *distal myodystrophies*
 - 3) *myotonic dystrophy*
 - 4) *debranching enzyme deficiency* (type III glycogenosis)
 - 5) *some families* with congenital myopathies
3. **SCAPULOPERONEAL weakness** - periscapular (proximal upper extremity) muscles and distal lower extremity anterior compartment muscles; etiology:
 - 1) *facioscapulohumeral dystrophy* (+ facial weakness)
 - 2) *scapuloperoneal syndromes*
 - 3) *Emery-Dreifuss muscular dystrophy* (humero-peroneal)
 - 4) *acid maltase deficiency*
 - 5) *some families* with congenital myopathies
4. **FOREARM** (wrist and finger flexors) + **THIGH** (quadriceps) **weakness** - pathognomonic for *inclusion body myositis* (weakness is often asymmetrical!).
5. **OCULAR** (ptosis, ophthalmoplegia without diplopia) + **PHARYNGEAL weakness**; etiology:
 - 1) *oculopharyngeal dystrophy*
 - 2) *mitochondrial myopathies* (without prominent pharyngeal involvement)
 - 3) *myotonic dystrophy* (ophthalmoplegia and pharyngeal involvement not in all cases)
 - 4) *myasthenia gravis* (ophthalmoplegia with diplopia)
6. **NECK EXTENSOR weakness** ("dropped head syndrome"); neck flexors may or may not be weak; etiology:
 - 1) amyotrophic lateral sclerosis
 - 2) myasthenia gravis
 - 3) polymyositis, dermatomyositis, inclusion body myositis

DIAGNOSTIC EVALUATION

1. **Electrodiagnosis** - EMG, nerve conduction studies, repetitive stimulation (abnormal in **neuromuscular junction disorders**) see p. D20 >>
2. Serum **CK**↑ (in **myogenic disorders**) see p. D30 >>
N.B. absence of CK↑ does not rule out myopathy (esp. severe muscle atrophy)!
3. Muscle / nerve **biopsy** (both normal in **neuromuscular junction disorders**) see p. D30 >>
4. **Cardiac evaluation** (ECG, etc) - heart involvement (cardiomyopathy, conduction defects) in **myogenic disorders**.
5. Serial **pulmonary function tests** in progressive diseases.

BIBLIOGRAPHY for ch. "Neuromuscular, Muscular Disorders" → follow this [LINK](#) >>