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**!

1. **Hypo**(**a**)**reflexia**
2. **Fasciculations** (specific for **neurogenic disorders**) [see p. Mov3 >>](http://www.neurosurgeryresident.net/Mov.%20Movement%20disorders%2C%20Ataxias%5CMov3.%20GENERAL%20-%20UMN%20%28pyramidal%29%20%26%20LMN%20Disorders.pdf)
3. **Sensory** changes (specific for **neurogenic disorders**)
4. **Myotonia** (specific for **myogenic disorders**); may be accompanied by muscle hypertrophy.
5. Painful **cramps** (esp. in **LMN disorders**; not in **neuromuscular junction disorders**).

[see p. Mov3 >>](http://www.neurosurgeryresident.net/Mov.%20Movement%20disorders%2C%20Ataxias%5CMov3.%20GENERAL%20-%20UMN%20%28pyramidal%29%20%26%20LMN%20Disorders.pdf)

1. **Myalgias**
2. **Muscle contractures** (**glycolytic enzyme defects**) - last longer than cramps, provoked by exercise, electrically silent. [see p. Mov3 >>](http://www.neurosurgeryresident.net/Mov.%20Movement%20disorders%2C%20Ataxias%5CMov3.%20GENERAL%20-%20UMN%20%28pyramidal%29%20%26%20LMN%20Disorders.pdf)
3. Tendon **contractures** (in **myopathies** of long duration; early contractures - Emery-Dreifuss dystrophy, Bethlem myopathy).
4. 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:
4. *facioscapulohumeral dystrophy* (+ facial weakness)
5. *scapuloperoneal syndromes*
6. *Emery-Dreifuss muscular dystrophy* (humeroperoneal)
7. *acid maltase deficiency*
8. *some families* with congenital myopathies
9. **Forearm** (wrist and finger flexors) **+ thigh** (quadriceps) **weakness** - pathognomonic for *inclusion body myositis* (weakness is often asymmetrical!).
10. **Ocular** (ptosis, ophthalmoplegia without diplopia) **+ pharyngeal** **weakness**; etiology:
11. *oculopharyngeal dystrophy*
12. *mitochondrial myopathies* (without prominent pharyngeal involvement)
13. *myotonic dystrophy* (ophthalmoplegia and pharyngeal involvement not in all cases)
14. *myasthenia gravis* (ophthalmoplegia with diplopia)
15. **Neck extensor** **weakness** ("dropped head syndrome"); neck flexors may or may not be weak; etiology:
16. amyotrophic lateral sclerosis
17. myasthenia gravis
18. polymyositis, dermatomyositis, inclusion body myositis

Diagnostic Evaluation

1. **Electrodiagnosis** - EMG, nerve conduction studies, repetitive stimulation (abnormal in **neuromuscular junction disorders**) [see p. D20 >>](http://www.neurosurgeryresident.net/D.%20Diagnostics%5CD20-29.%20Electrophysiology%20%28EEG%2C%20evoked%20potentials%2C%20MEG%2C%20EMG%2C%20nerve%20conduction%29%5CD20.%20EMG.pdf)
2. Serum **CK↑** (in **myogenic disorders**) [see p. D30 >>](http://www.neurosurgeryresident.net/D.%20Diagnostics%5CD30-39.%20Biopsy%20%28brain%2C%20nerve%2C%20muscle%29%5CD30.%20Muscle%20Biopsy%20and%20Serum%20Markers.pdf)

N.B. absence of CK↑ does not rule out myopathy (esp. severe muscle atrophy)!

1. Muscle / nerve **biopsy** (both normal in **neuromuscular junction disorders**) [see p. D30 >>](http://www.neurosurgeryresident.net/D.%20Diagnostics%5CD30-39.%20Biopsy%20%28brain%2C%20nerve%2C%20muscle%29%5CD30.%20Muscle%20Biopsy%20and%20Serum%20Markers.pdf)
2. **Cardiac evaluation** (ECG, etc) - heart involvement (cardiomyopathy, conduction defects) in **myogenic disorders**.
3. Serial **pulmonary function tests** in progressive diseases.

Bibliography for ch. “Neuromuscular, Muscular Disorders” → follow this [link >>](http://www.neurosurgeryresident.net/Mus.%20Muscular%2C%20Neuromuscular%20disorders%5CMus.%20Bibliography.pdf)

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