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* (humeroperoneal)
   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 >>