Neuromuscular Disorders (GENERAL)

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CLINICAL MANIFESTATIONS

1. Symmetrical flaccid muscle weakness
2. Muscle wasting \(\text{or} \) atrophy \(\text{or} \) pseudohypertrophy
   - Advisable to use term “wasting” in myogenic disorders and “atrophy” in neurogenic disorders!
3. Hyporeflexia
4. Fasciculations \(\text{specific for neurogenic disorders}\) see p. Mov3 >>
5. Sensory changes \(\text{specific for neurogenic disorders}\)
6. Myotonia \(\text{specific for myogenic disorders}\); may be accompanied by muscle hypertrophy.
7. Painful cramps \(\text{esp. in LNM disorders; not in neuromuscular junction disorders}\).

Diagnosis Evaluation of the following:

1. Blood tests
2. Genetic analysis
3. Electromyography
4. Muscle biopsy
5. Imaging studies

CLINICAL MANIFESTATIONS

1. Lower motor neuron / its axon – neurogenic disorders
2. Myogenic disorders
3. Neuromuscular junction - neuromuscular junction disorders
4. Muscle fibers – myogenic disorders

NEUROGENIC disorders:

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

NEUROMUSCULAR JUNCTION disorders:

1. variable fatigable weakness \(\text{initially in extraocular \& bulbar muscles}\)
2. no atrophy!!!
3. reflexes present!!!
4. no fasciculations, no sensory loss.

MYOGENIC disorders:

1. initially affects large proximal muscle groups \(\text{with exceptions – see below}\); neck flexion is much weaker than neck extension.
2. weakness \(\rightarrow\) atrophy
3. reflexes long present \(\text{diminished in proportion to weakness degree}\)
4. no fasciculations, no sensory loss
5. possible myalgias \(\text{surprisingly uncommon in most muscle diseases!}\), 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 subcutaneous ribs \(\text{due to intercostal muscle weakness}\).
- male infants may have undescended testicles \(\text{due to weak gubernaculum}\).
- developmental delay.

Try to establish pattern of weakness:

1. LIMB-GERI weakness \(\text{most common and therefore least specific pattern}\) - weakness exclusively / predominantly in limb proximal muscles; neck flexor \& extensor muscles can also be affected. – etiology – 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. neurosopathies \(\text{most commonly}\)
     2. distal myosopathies
     3. myotonic dystrophy
     4. debranching enzyme deficiency \(\text{type III glycosogen}\)
     5. some families with congenital myopathies
3. SCAPULOPHRENAL weakness - periscapular \(\text{proximal upper extremity}\) muscles and distal lower extremity anterior compartment muscles; etiology:
   1. facioscapulohumeral dystrophy \(\text{\& facial weakness}\)
   2. scapuloperoneal syndrome
   3. Emery-Dreifuss muscular dystrophy \(\text{humeroperoneal}\)
   4. acid maltase deficiency
   5. some families with congenital myopathies
4. FOREARM \(\text{wrist and finger flexors} +\) THIGH (quadriiceps) weakness - pathognomonic for inclusion body myositis \(\text{weakness is often asymmetrical}\).
5. OCULAR \(\text{pseudohyphalmoplegia without diplopia} +\) PSEUDOHYPOGLAN weakness; etiology:
   1. oculopharyngeal dystrophy
   2. mitochondrial myopathies \(\text{without prominent pharyngeal involvement}\)
   3. myotonic dystrophy \(\text{ophthalmoplegia and pharyngeal involvement not in all cases}\)
   4. myasthenia gravis \(\text{ophthalmoplegia with diplopia}\)
6. NECK EXTENSOR weakness \(\text{\& dropped head syndrome}\); neck flexors may or may not be weak.
   - amyotrophic lateral sclerosis
   - myasthenia gravis
   - 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)  
   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 >>