# PART I. INDIVIDUAL NEUROMUSCULAR DISORDERS

# 1. MOTOR NEURONOPATHIES

## AMYOTROPHIC LATERALS SCLEROSIS (ALS)

### Etiology

- heterogeneous etiology
- most are sporadic
- 5-10 % are hereditary, both dominant and recessive inheritance has been described:
- one hereditary form is linked to chromosome 2q33-q35, another is Cu/Zn superoxide dismutase (SOD1) mutation on chromosome 21q22
- degeneration of both upper and lower motor neurons

### Clinical features

- incidence around 2/100 000
- usually onset in the older age groups around 60 years but may present earlier
- males>females
- painless weakness
- upper limb muscles affected more frequently than lower limb muscles
- bulbar muscles may be involved; slurring of speech, dysphagia,
- extraocular muscles and anal or urethral sphincters are rarely clinically affected
- upper motor neuron signs, increased tendon reflexes and positive Babinski sign, often present
- average survival after diagnosis 3 years,;in patients presenting with bulbar symptoms shorter survival

### Strategy

- confirm neurogenic EMG findings in muscles in three regions of the body (the regions are (1) brainstem, (2) brachial, (3) thorax, (4) trunk and (5) crural
- exclude polyneuropathy
- differentiate from multifocal motor neuropathy with conduction block (MMN)
- differentiate from spinal hereditary motor neuronopathies (spinal muscular atrophies)
- differentiate from previous polio

### Diagnostic criteria

- The diagnosis of ALS requires the presence of: (1) lower motor neuron signs (LMS), (2) upper motor neuron signs (UMS) and (3) progression of the disorder
    1. *definite ALS*: LMS in three regions and UMS
    2. *probable ALS*: LMS in two regions and UMS
    3. *possible ALS:* LMS in one region and UMS
    4. *suspected ALS:* LMS in two or three regions

### Expected abnormal findings

*EMG*

- subacute neurogenic muscle findings in at least 3 regions. Preferentially the findings should be asymmetric without any definite proximal or distal predominance.

*Neurography*

- MCS often show reduced amplitudes
- If the AMPL is reduced significantly the conduction velocity may be reduced (loss of fastest conducting axons)

*Central motor conduction time*

- often abnormal

### Expected normal findings

*Neurography*

- SCS
- MCS: if AMPL is normal or only moderately reduced, CV should be normal
- no motor conduction block
- normal number of F-waves in mildly affected muscles (indicating lack of proximal conduction block)

*Evoked potentials*

- VEP
- BAEP
- SEP (may rarley be abnormal)

### Procedure

*EMG (if normal findings on one side examine both sides!)*

- m.orbicularis oris/m.masseter/m.genioglossus (insert electrode below chin)
- m.trapezius
- interosseus dorsalis I / m.biceps brachii / m.deltoideus
- m.tibialis anterior / m.gastrocnemius caput mediale / m.vastus lateralis
- if the patient presents with dysarthria and there are no other EMG abnormalities, test m.chricothyreoideus

*Neurography MCS (bilaterally)*

- n.medianus
- n.ulnaris (also including supraclavicular stimulation)
- n.peroneus
- n.tibialis

*Neurography SCS (bilaterally)*

- n.suralis
- n.radialis

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**Modified**
" BF 25.3.1997 ES 2.4.97

## POLIO, ACUTE

**Etiology**
" following infection by poliovirus most patients will have fever and gastrointestinal symptoms, but 1-2 % develope paralysis
" due to vaccination polio has been eradicated from developed countries
" attenuated virus used for vaccination may cause paralysis, estimated risk is 1 per million
" sometimes coxackie and eccho virus may cause a similar paralysis

**Clinical features**
" around 1-2 weeks after an episode of febrile illness 1-2 % of patients with a polio virus infection will develop a paralytic disorder
" the risk of paralysis increases with age, adults are about 10 times as likely to develop paralysis than children
" any group of motor neurons may be affected, the distribution of weakness varies
" after the initial paralysis a varying degree of restitution takes place
" some patients with polio perceive after a stable period of more than 15 years new loss of muscle function (often called post-polio syndrome, see *Polio, sequale following previous infection*

**Strategy**
" show acute neurogenic involvement of several muscles usually in several regions of the body
" assess distribution of abnormalities and degree of involvement
" differentiate from polyradiculitis

**Expected abnormal findings**
*EMG*
" acute neurogenic EMG findings in muscles
" distribution not confined to one segment or nerve; if local - reconsider diagnosis
*Neurography*
" MCS: reduced amplitudes in weak muscles
" if the amplitude is severely reduced the MCV may be reduced

**Expected normal findings**
*Neurography*
" SCS

**Procedure**
*EMG*
" weak muscles should be studied
*Neurography*
The following motor nerves should be studied bilaterally
" n.medianus
" n.ulnaris
" n.peroneus
" n.tibialis
The following sensory nerves should be tested
" n.suralis
" n.radialis

**Note**
" although vaccination has largely eradicated acute polio from countries with a good vaccination program, polio may still occur in some parts of the world. In industrialized countries there are minorities that are negative to vaccinations and refugees that may still lack vaccination against polio.

**References**


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" 2.4.1997, ES 3.4.97, BF 15.4.1997

## POLIO, SEQUALE FOLLOWING PREVIOUS PARALYSIS

**Etiology**

" previously suffered from polio, the patient has recovered from the acute episode to various degrees
" the causes of sequele vary considerably: (1) normal aging with loss of contractile strength contributesloss of motor units with aging, (2) pain from joints and tendons and (3) psycho-social factors
" there are no well defined biological parameters that define this disorder

**Clinical features**

" after a stable period of more than 15 years after the initial paralysis some patients develop subjective new loss of muscle function (often called post-polio syndrome)
" the distribution of weakness varies considerably and depends on the distribution and severity of the initial affection

**Strategy**

" show inactive neurogenic involvement of several muscles usually in several regions of the body
" make sure the polio diagnosis is correct
" assess distribution of abnormalities and degree of involvement
" beware of additional diseases: radiculopathy, CTS, polyneuropathy, polymyositis

**Expected abnormal findings**

*EMG*

" inactive neurogenic findings in weak and often also in strong muscles depending on involvement
" distribution not confined to one segment or nerve (if findings are focal - reconsider diagnosis)

*Neurography*

" MCS may show reduced amplitudes and if the amplitude is severely reduced the MCV may be reduced

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

" the muscles studied depends entirely on the clinical symptoms..
" if the diagnosis is obvious, neurography is not necessary, unless the patient has a clinical problem that warrants neurography.
" Macro EMG for quantitation of motor unit size
" motor unit counting for quantification of number of motor units

**Note**

" neurophysiological findings or other laboratory findings do not differentiate between patients that are stabe and those that experience new loss of muscle function (post-polio syndrome)

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" 2.4.1997 BF, ES:3.4.97

## SPINAL MUSCULAR ATROPHY 1, WERDNIG-HOFFMAN

**Etiology**

" autosomal recessive inheritance
" genetic defect localized to chromosome 5q11-q13
" deletion in a region that codes the SMN (the survival motor neuron gene) and the NAIP (neuronal apoptosis inhibitory protein)
" good correlation between the size of the deletion and the severity of the phenotype
" incidence of deletions of both SMN and NAIP is 62% in SMA type I, 8.8% in type II and 12% in type III
" degeneration of the anterior horn cells in the spinal cord

**Clinical features**

" second most common lethal autosomal recessive disorder

- onset in utero or before the age of 3 months
- hypotonia and weakness, often the child is in a frog posture
- respiratory problems, diaphragmatic breathing, costal recession
- absent tendon reflexes
- intellectually normal
- poor prognosis; most die within the first year
- CK normal or moderately elevated
- Incidence is around 1/100000 (1/67000-1/25000 has been reported)
- carrier rate of the gene in the general population is around 1/80

**Strategy**

- demonstrate acute or subacute neurogenic EMG findings of limb muscles
- demonstrate normal sensory neurography

**Expected abnormal findings**

*EMG*

- acute or subacute neurogenic EMG findings in several muscles (proximal muscles tend to be more involved than distal muscles)

*Neurography*

- MCS: reduced AMPL, CV may be reduced if AMPL is very low

**Expected normal findings**

*Neurography*

- SCS

**Procedure**

*EMG (unilaterally)*

- m.deltoideus/m.biceps brachii
- m.interosseus dorsalis
- m.vastus lateralis
- m.tibialis anterior

*Neurography*

- one motor and sensory nerve in the upper and lower extremities

**Note**

- sometimes the muscles shows typical continuous involuntary motor unit activity

**References**


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- 26.3.1997 BF, ES 3.4.97, 8.5.1997 BF

## SPINAL MUSCULAR ATROPHY 2, INTERMEDIATE

**Etiology**

- autosomal recessive inheritance
- genetic defect localized to chromosome 5q11-q13
- deletion in a region that codes the SMN (the survival motor neuron gene) and the NAIP (neuronal apoptosis inhibitory protein).
- degeneration of the anterior horn cells in the spinal cord
- good correlation between the size of the deletion and the severity of the phenotype
- incidence of deletions of both SMN and NAIP is 62% in SMA type I, 8.8% in type II and 12% in type III.
- degeneration of the anterior horn cells in the spinal cord

**Clinical features**

- onset between 6 and 12 months,
- symmetric weakness of proximal weakness, more in the legs than arms
- never learn to walk (in contrast to patients with SMA 3 who will learn to walk)
- tendon reflexes absent
- facial muscles are clinically spared
- scoliosis
- normal intellect
- slowly progressive
- CK: normal or moderately elevated

**Strategy**

- demonstrate chronic neurogenic EMG findings of limb muscles (proximal muscles more involved than distal muscles)
- demonstrate normal sensory neurography

**Expected abnormal findings**

*EMG*

- acute or subacute neurogenic EMG findings in several muscles

*Neurography*

- MCS: reduced AMPL, CV may be reduced if AMPL is very low

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG (unilaterally)*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" one motor and sensory nerve in the upper and lower extremities

**References**


" Burlet P, Burglen L, Clermont O, et al. Large scale deletions of the 5q13 region are specific to Werdnig-Hoffmann disease. J Med Genet 1996;33:281-283
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**Modified**

" 26.3.1997 BF ES 3.4.97

## SPINAL MUSCULAR ATROPHY 3, KUGELBERG-WELANDER

**Etiology**

" autosomal recessive inheritance
" genetic defect localized to chromosome 5q11-q13
" deletion in a region that codes the SMN (the survival motor neuron gene) and the NAIP (neuronal apoptosis inhibitory protein).
" degeneration of the anterior horn cells in the spinal cord
" good correlation between the size of the deletion and the severity of the phenotype
" incidence of deletions of both SMN and NAIP is 62% in SMA type I, 8.8% in type II and 12% in type III.
" degeneration of the anterior horn cells in the spinal cord

**Clinical features**

" onset between 1-20 years of age
" learn to walk but loose later the ability to walk
" present with difficulties with walking, running climbing or jumping
" Gower's sign
" proximal weakness
" legs weaker than arms
" hypermobility of joints
" CK normal or moderately elevated
" slow progression

**Strategy**

" demonstrate symmetric chronic neurogenic EMG findings of limb muscles (proximal muscles more involved than distal muscles)
" demonstrate normal sensory neurography

**Expected abnormal findings**

*EMG*

" acute or subacute neurogenic EMG findings in several muscles

*Neurography*

" MCS: reduced AMPL, CV may be reduced if AMPL is very low

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG (unilaterally)*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" one motor and sensory nerve in the upper and lower extremities

**References**


" Burlet P, Burglen L, Clermont O, et al. Large scale deletions of the 5q13 region are specific to Werdnig-Hoffmann disease. J Med Genet 1996;33:281-283
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## X-LINKED BULBOSPINAL HEREDITARY NEURONOPATHY (KENNEDY SYNDROME)

**Etiology**
" X-linked recessive inheritance
" Xq21-22, mutation consists of expansion of CAG tinucleotide repeats (disease severity correlated with number of CAG repeats)
" gene product: androgen receptor
" spinobulbar motor neuronopathy

**Clinical features**
" onset at the age 30 to 50 years, sometimes as young as 15 years
" muscle cramps often initial symptom
" proximal muscle weakness, legs > arms
" facial and bulbar muscles affected
" distribution of affected muscles in the beginning varies somewhat, patients often complain about focal weakness.
" gynecomastia, impotence, infertility
" absent of depressed tendon reflexes
" normal life expectancy
" CK normal or mildly elevated (up to five times normal)

**Strategy**
" demonstrate subacute neurogenic abnormalities with a proximal and symmetric distribution
" differentiate from sensory-motor polyneuropathy
" differentiate from ALS
" differentiate from previous polio

**Expected abnormal findings**
*EMG*
" symmetric subacute neurogenic abnormalities with proximal distribution, fasciculations common
*Neurography*
" mild sensory abnormalities with low sensory amplitudes

**Procedure**
*EMG (unilaterally)*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis
" m.vastus lateralis
" m.tibialis anterior
*Neurography*
" one motor and sensory nerve in the upper and lower extremities

**References**
" Kennedy WR, Alter M, Sung J H. Progressive proximal spinal and bulbar muscular atrophy of late onset: a sex-linked recessive trait. Neurology 1968;18: 671-680
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## DISTAL SPINAL MUSCULAR ATROPHY

**Etiology**
" genetically heterogeneous
" there are several autosomal dominant forms and recessive forms
" degeneration of spinal anterior horn cells

**Clinical features**
" patients with dominant form of the disease develop symptoms during the firs decade, usually before 20 years of age
" distal weakness and wasting of muscles
" often pes cavus
" legs more affected than arms
" tendon reflexes normal or depressed

**Strategy**
" demonstrate subacute neurogenic abnormalities in distal limb muscles
" differentiate from sensory-motor polyneuropathy and ALS

**Expected abnormal findings**

*EMG*

" subacute neurogenic abnormalities in distal muscles bilaterally
" proximal muscles less affected

*Neurography*

" MCS low AMPL

**Expected normal findings**

*Neurography*

" normal SCS

**Procedure**

*EMG (unilaterally)*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis
" m.vastus lateralis
" m.tibialis anterior

*Neurography (unilaterally)*

" MCS: n.medianus, n.peroneus
" SCS: n.suralis, n.radialis

**References**


" Boylan KB, Cornblath DR, Glass JD, Alderson K, Kuncl RW, Kleyn PW, Gilliam TC. Autosomal dominant distal spinal muscular atrophy in four generations. Neurology 1995;45:699-704
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**Modified**

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## MONOMELIC SPINAL MUSCULAR ATROPHY

**Etiology**

" unknown
" most are sporadic
" autosomal dominant inheritance has been described

**Clinical features**

" onset usually at the age of around 20 years, mostly before the age of 30
" weakness and wasting of intrinsic hand muscles, forearm muscles may be affected
" progression lasts usually 1-2 years and then the process is arrested
" cases affecting proximal leg muscles have been described

**Strategy**

" demonstrate focal subacute neurogenic abnormalities within a few myotomes, usually C7-C8-Th1
" findings are usually bilateral
" differentiate from ALS and MMN

**Expected abnormal findings**

*EMG*

" subacute neurogenic abnormalities in C7-C8-Th1 muscles

*Neurography*

" MCS low AMPL in ulnar, median and radial nerves

**Expected normal findings**

*Neurography*

" normal SCS

*EMG*

" normal findings in leg muscles and muscles innervated by cranial nerves

**Procedure**

*EMG*

" m.deltoideus
" m.biceps
" m.triceps, bilaterally
" m.extensor digitorum communis, bilaterally
" m.interosseus dorsalis, bilaterally
" m.opponens pollicis/m.abductor pollicis brevis, bilaterally
" m.vastus lateralis
" m.tibialis anterior
" m.trapezius

*Neurography*

" n.medianus, MCS and SCS
" n.ulnaris, MCS and SCS
" n.radialis, SCS
" n.suralis, SCS
" n.peroneus, MCS

**References**


" De Visser M, de Visser BW, Verbeeten B. Electromyographic and computed tomographic findings in five patients with monomelic spinal muscular atrophy. Eur Neurol 1985; 28:135-138

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### BULBAR HEREDITARY MOTOR NEURONOPATHY (FAZIO-LONDE'S DISEASE)

**Etiology**
" unknown motor neuronopathy of the bulbar motor neurons

**Clinical features**
" onset usually at 2 to 3 years age, latest 12 years
" dysphagia
" facial weakness
" extraocular palsies
" limb muscles may be involved
" most patients die within two years of onset

**Strategy**
" demonstrate motor neurogenic EMG findings of muscles innervated by the cranial nerves
" differentiate from SMA

**Expected abnormal findings**
*EMG*
" subacute neurogenic abnormalities in muscles innervated by the cranial nerves
" limb muscles may be involved
*Neurography*
" MCS low AMPL in n.facialis

**Expected normal findings**
*Neurography*
" normal SCS

**Procedure**
*EMG*
" m.orbicularis oris
" m.masseter
" m.trapezius
" m.deltoideus/m.biceps
" m.interosseus dorsalis/m.opponens pollicis/m.abductor pollicis brevis
" m.vastus lateralis
" m.tibilalis anterior
*Neurography*
" n.medianus, MCS and SCS
" n.suralis, SCS
" n.peroneus, MCS

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**Modified**
" 26.3.1997 BF, 3.4.97

# 2. POLYNEUROPATHIES

### AXONAL SENSORY-MOTOR POLYNEUROPATHY

**Etiology**
" almost 200 different metabolic, toxic and genetic causes are known
" most common cause in the developed countries is diabetes
" other common causes are renal insufficiency and chronic alcoholism

**Clinical features**
" the sensory and motor symptoms vary considerably and depend to some extent on the etiology
" weakness of distal muscles, legs affected more than arms
" sensory loss starting in the distal part of the legs and spreading proximally
" paresthesias, dysesthesias in the distal parts of the extremities usually worse at rest
" muscle cramps

**Strategy**
" demonstrate generalized axonal dysfunction of peripheral sensory and motor nerves
" often also autonomic nerves are affected to varying degrees.
" are the findings compatible with a specific axonal polyneuropathy?
*Assess*
" type (motor/sensory/autonomic)
" time course (acute, subacute, inactive)
" severity

" pathophysiology (axonal/demyelinating/conduction block)
" distribution: diffuse (distal/proximal) or multifocal

**Expected abnormal findings**

*Neurography*
" SCS ampl reduced. Distal leg nerves are more affected than arm nerves.
" in moderate to severe axonal polyneuropathies also motor amplitudes are reduced
" CV is slow normal or slightly reduced (not more than 30% of reference values) in axonal polyneuropathies
" F waves are delayed and the number of F waves may be reduced
" A-waves are often seen

*EMG*
" limb muscles show varying degrees of neurogenic involvement. Distal muscles are more affected than proximal muscles. Leg muscles are more affected than arm muscles

*Autonomic testing*
" often abnormal

*Sensory thresholds*
" often abnormal

**Procedure**

*Neurography*
" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.

*EMG (optional)*
" one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*
" temperature and vibration thresholds

## DEMYELINATING SENSORY-MOTOR POLYNEUROPATHY

**Etiology**
" various causes have been described. Demyelinating polyneuropathies are much less common than axonal polyneuropathies
" diphtheria
" monoclonal gammopathies and paraneoplastic
" inflammatory: acute polyradiculitis, chronic polyradiculitis
" hereditary: HMSN1, HMSN3, HMSN4, HNPP

**Clinical features**
" the sensory and motor symptoms vary considerably and depend to some extent on the etiology
" weakness of distal muscles, legs affected more than arms
" sensory loss starting in the distal part of the legs and spreading proximally
" paresthesias, dysesthesias in the distal parts of the extremities usually worse at rest
" muscle cramps

**Strategy**
" demonstrate generalized demyelinating dysfunction of peripheral sensory and motor nerves.
" are the findings compatible with a specific demyelinating polyneuropathy? (polyradiculitis, HMSN1, HNPP)

*Assess*
" type (motor/sensory/autonomic)
" time course (acute, subacute, inactive)
" severity
" pathophysiology (axonal/demyelinating/conduction block)
" distribution: diffuse (distal/proximal) or multifocal

**Expected abnormal findings**

*Neurography*
" sensory and motor nerve conduction velocities are reduced, usually by more than 30%
" distal motor latencies often > 7 ms
" the amplitude of the sensory nerve action potentials are reduced. Distal leg nerves are more affected than arm nerves.
" in severe demyelinating axonal polyneuropathies also motor amplitudes are reduced due to secondary axonal involvement
" F wave latencies are delayed and the number of F waves may be redduced
" A-waves may be seen

*EMG*
" limb muscles show varying degrees of neurogenic involvement. distal muscles are more affected than proximal muscles. leg muscles are more affected than arm muscles

*Autonomic testing*
" often abnormal

*Sensory thresholds*
" often abnormal

**Procedure**

*Neurography*
" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally

*EMG (optional)*
" one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

**Note**

" hereditary demyelinating polyneuropathies usually do not display conduction block
" acquired demyelinating polyneuropathies tend to have conduction block

## ACUTE POLYRADICULITIS (GUILLAIN-BARRÉ SYNDROME, GBS, ACUTE INFLAMMATORY DEMYELINATING POLYNEUROPATHY, AIDP)

**Etiology**

" probably autoimmune reaction against peripheral nerves sometimes triggered by preceding infection (especially Campylobacter jejuni) trauma, operation or childbirth.

**Clinical features**

" incidence 1-2/100000 per year in people younger than 45 years, in 70-75 year old the incidence is 4-6/100000
" male : female ratio 1,5:1
" occurs in all age groups, peak 50-70 years
" acute onset within days
" at the onset 80 % have paresthesias and 60 % have weakness
" paresthesias precede the weakness by a few days
" typically patients notice weakness in the legs before the arms
" the presentation of weakness is often ascending (50-60%) from legs to arms and cranial muscles
" the presentation may be descending in a portion of patients
" pain is not uncommon; 15-50 % of patients have pain
" facial nerve is involved in 50 %
" the autonomic nervous system may be affected, especially in patients with severe motor deficits
" tendon reflexes are decreased or absent
" most patients worsen over 1-2 weeks, some for up to 4 weeks
" mortality2-5 %
" after plateau subsequent recovery over 6-12 months
" patients that worsen for more than 8 weeks probably have chronic polyradiculitis
" classically it has been thought that polyradiculatis is primarily a demyelinating sensory and motor polyneuropathy, currently it is accepted that there is an primarily axonal form of polyradiculitis, sometimes affecting only motor nerves (acute motor axonal neuropathy AMAN)

**Strategy**

" demonstrate acute motor and sensory neuropathy
" in the acute stage the motor nerves are more affected than sensory nerves
" the neuropathy is typically demyelinating with conduction block, the most prominent findings are often in the proximal parts of the nerves
" sometimes the neuropathy is predominantly axonal, especially if associated with Campylobacter jejuni
" assess: severity, pathology, distribution

**Expected abnormal findings**

*Neurography, MCS*

" conduction block (dist/prox amplitude >30 % in upper extremities and > 50 % in lower extremities without considerable increase in dispersion)
" F waves delayed and few due to conduction block
" DL prolonged
" reduced MCV, sometimes normal initially
" distal amplitude may be initially normal - low amplitude with normal DL indicates severe axonal involvement

*Neurography SCS*

" reduced SCV is not necessarily seen during the first weeks
" reduced amplitude is seen in the presence of axonal loss

*EMG*

" < 10-18 days from onset of symptoms: only reduced interference pattern
" > 10-18 days from onset: signs of acute neurogenic EMG findings

*Autonomic tests*

" often abnormal

**Procedure**

*Neurography*

" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.radialis, n.suralis unilaterally.

*EMG*

" < 10 days from onset of symptoms not necessarily informative, but may reveal earlier onset and confirm peripheral cause of weakness
" > 10 days from onset, should be done to demonstrate degree of axonal involvement.

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

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**Modified**

" 25.6.1997 BF, 2.4.1997 BF 3.4.97 ES

## ACUTE AXONAL MOTOR NEUROPATHY (AMAN)

**Etiology**

" immune reaction against motor axons, probably triggered by preceding infection, especially Campylobacter jejuni, trauma, operation or childbirth.

**Clinical features**

" most patients described come from northern China and Mexico
" usually triggered by Campylobacter jejuni infection
" most patients are children or young adults
" most cases occur during summer months, peak in augus
" rapidly developing symmetric flaccid paralysis, may progress to tetraparesis and respiratory failure
" extraocular mucles rarely involved
" mortality around 5 %
" CSF shows elevation of proteins during secopnd or third week from onset
" prognosis is usually favourable in spite of the axonal involvement, it is thought that the axonal involvement is distal
" the prognosis does not differ from the classical demyelinating sensory-motor type of polyradiculitis

**Strategy**

" demonstrate acute motor axonal neuropathy
" differentiate from polio, which tends to be asymmetricac and pathcy
" assess: severity, pathology, distribution

**Expected abnormal findings**

*Neurography, MCS*

" DL prolonged
" mildly reduced MCV, sometimes normal initially
" M wave amplitude reduced
" F waves absent or number reduced

*EMG*

" < 10-18 days from onset of symptoms: only reduced interference pattern
" > 10-18 days from onset: signs of acute neurogenic EMG findings

**Expected normal findings**

*Neurography, SCS*

" all sensory nerves should be normal

*Sensory thresholds*

" temperature and vibration

**Procedure**

*Neurography*

" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.radialis, n.suralis unilaterally.

*EMG*

" < 10 days from onset of symptoms not necessarily informative, but may reveal earlier onset and confirm peripheral cause of weakness
" > 10 days from onset, should be done to demonstrate degree of axonal involvement.

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

**References**


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**Modified**

" 25.6.1997 BF, 2.4.1997 BF 3.4.97 ES

## MILLER-FISHER SYNDROME

### Etiology

" probably autoimmune reaction against peripheral nerves sometimes triggered by preceding infection (particularly Campylobacter jejuni) trauma, operation or childbirth
" considered to be a variant of acute polyradiculitis

### Clinical features

" characterized by external ophthalmoplegia, ataxia and areflexia
" patients experience diplopia and unsteadiness of gait, either of these symptoms may be the initila sign
" other muscles innervated by cranial nerves may show weakness; dysarthria, facial palsy, ptosistongue weaknsess, palatal palsy
" acute onset within days, peak of symptoms is reached around one to two weeks from onset
" mild weakness and sensory symptoms may be present
" usually the course is monophasic and relatively benign, recurrences have been described
" CSF protein is elevated

### Strategy

" demonstrate acute motor and sensory neuropathy
" in the limbs the neuropathy is predominantly sensory
" assess: severity, pathology, distribution

### Expected abnormal findings

*Neurography, MCS*

" mild slowing of motor CV
" F waves delayed and few due to conduction block
" DL may be prolonged
" mildly reduced MCV, sometimes normal initially
" distal amplitude reduced, especiall in facial muscles

*Neurography SCS*

" reduced amplitude dominates over reduced CV

*EMG*

" < 10-18 days from onset of symptoms: only reduced interference pattern
" > 10-18 days from onset: signs of acute neurogenic EMG findings

*Autonomic tests*

" often abnormal

*Blink reflex*

" absent or mildly prolonged R1 components
" R2 may be absent, but latency is usually normal

### Procedure

*Neurography*

" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" MCS: the branches of n.facialis bilaterally
" SCS: n.radialis, n.suralis unilaterally.

*EMG*

" < 10 days from onset of symptoms not necessarily informative, but may reveal earlier onset and confirm peripheral cause of weakness
" > 10 days from onset, should be done to demonstrate degree of axonal involvement.
" facial muscles should be studied

*Blink reflex*

" if supraorbital nerve stimulation gives normal findings, consider doing also the infraorbital nerve

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

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**Modified**
" 25.6.1997 BF

## CHRONIC POLYRADICULITIS (CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY, CIDP)

**Etiology**
" probably prolonged autoimmune reaction against peripheral nerves.

**Clinical features**
" occurs in all age groups from 2 years
" may begin as a typical acute polyradiculitis but may start with subacute symptoms as well
" either chronic progressive (slowly or stepwise) course or relapsing
" progression > 6 weeks followed by episodes of improvement and worsening for at least 3 months or slow progression for > 6 months
" bilateral relatively symmetric weakness
" paresthesias in toes and fingers
" facial muscles may be affected (10-15% of patients)
" elevated CSF protein concentration during deterioration
" areflexia at ankles and general hyporeflexia for at least 1 month
" patients with a relapsing course often resolve after a few years and they tend to have a better prognosis than those with chronic progressive course
" MRI shows increased signals on T2 weighted images at the sites of conduction blocks. MRI of the brachial plexus or other regions with suspected conduction blocks may be a useful accessory investigation in the diagnosis

**Strategy**
" demonstrate subacute motor and sensory neuropathy
" the neuropathy is typically demyelinating with conduction block, the most prominent findings are often in the proximal parts of the nerves
" sometimes the neuropathy is predominantly axonal
" assess: severity, pathology, distribution

**Expected abnormal findings**
*Neurography, MCS*
" reduced MCV
" conduction block (dist/prox amplitude >30 % in upper extremities and > 50 % in lower extremities without considerable increase in dispersion)
" F waves delayed and few due to conduction block
" dist lat prolonged
" distal ampl may be initially normal - low amplitude with normal dist.lat. indicates severe axonal involvement

*Neurography SCS*
" reduced SCV
" reduced amplitude is seen in the presence of axonal loss

*EMG*
" depends on severity of disorder, in mild cases distal muscles show subacute or inactive neurogenic findings in more severe cases also proximal muscles are involved

*Autonomic nervous system tests*
" RR-interval often abnormal
" SSR may be abnormal

**Procedure**
*Neurography*
" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.radialis, n.suralis unilaterally.

*EMG*
" one proximal and distal muscle in the upper and lower extremities

*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*
" temperature and vibration

**References**
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**Modified**
" 25.6.1997 BF, 2.4.1997 BF, 3.4.97 ES

## MULTIFOCAL MOTOR NEUROPATHY WITH CONDUCTION BLOCK (MMN)

**Etiology**
" unknown, possibly an autoimmune reaction against gangliosides ($GM_1$) in some patients
" regarded by many as a variant of CIDP

**Clinical features**

- slowly progressive weakness, usually distributed within one peripheral nerve (in ALS the distribution follows spinal myotomes)
- progression usually slow over years and decades
- weakness is often distally accentuated, but may be proximal in some patients
- muscle atrophy of some weak muscles is less pronounced than would be expected (weakness may be due to conduction block)
- fasciculations, cramps and myokymia may be seen
- MMN is predominantly a motor neuropathy, but mild sensory symptoms and findings may be seen
- areflexia may be seen in clinically not affected muscles
- no signs of upper motor neuron lesion
- very rarely involvement of cranial nerves
- m.diaphragma is rarely affected
- the resemblance of MMN to ALS; clinically it may sometimes be difficult to distinguish them
- differentiation of ALS and MMN can readily be done with EMG and nerve conduction studies
- MRI shows increased signals on T2 weighted images at the sites of conduction blocks. MRI of the brachial plexus or other regions with suspected conduction blocks may be a useful accessory investigation in the diagnosis of MMN.

## Strategy

- confirm subacute neurogenic EMG findings of muscles in several parts of the body
- demonstrate significant conduction block in motor nerves
- exclude sensory motor polyneuropathy
- differentiate from ALS
- differentiate from spinal hereditary motor neuronopathies (spinal muscular atrophies)
- differentiate from previous polio

## Expected abnormal findings

*EMG*

- subacute or chronic neurogenic muscle findings in several nerves (the weakness and EMG findings are often related with individual nerves rather than myotomes)

*Neurography*

- motor nerve show conduction block (amplitude and area decay, reduced number of F-waves)
- MCS often show reduced amplitudes
- motor conduction velocity may be reduced, especially if amplitude is small

## Expected normal findings

*Neurography*

- SCS normal

*Central motor conduction time*

- normal

## Procedure

*EMG*

The muscles should be chosen based on the clinical muscle weakness; if weakness is widespread, the following muscles are recommended

- m.interosseus dorsalis I
- m.biceps brachii/m.deltoideus
- m.tibialis anterior/m.gastrocnemius caput mediale
- m.vastus lateralis
- m.trapezius/orbicularis oris/m.genioglossus (insert electrode below chin)

*Neurography*

The following motor nerves should be studied bilaterally

- n.medianus
- n.ulnaris (also including supraclavicular stimulation)
- n.peroneus
- n.tibialis

The following sensory nerves should be tested

- n.suralis
- n.radialis

## Note

- patients with MMN do not have upper motor neuron signs
- muscles innervated by cranial nerves are rarely affected

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**Modified**

" 25.6.1997 BF, 31.3.1997, 2.4.1997

## HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 1 (HMSN1, CHARCOT-MARIE-TOOTH)

**Etiology**

" several independent subtypes have been described
" 1a is most common (70-95%). Autosomal dominant inheritance, linked to chromosome 17p11.2. Gene product peripheral myelin protein 22 (PNP-22)
" 1b is less common. Autosomal dominant inheritance, linked to chromosome 1q21-23. Gene product peripheral myelin protein $P_0$ (PNPO)
" 1c, other autosomal loci have been described
" X-linked dominant (X1), linked to Xq13, gene product connexin
" X-linked recessive (X2) have been described

**Clinical features**

" a relatively common polyneuropathy
" slowly developing sensory and motor polyneuropathy
" first symptoms usually during the second decade but may start earlier or considerably later
" peroneal muscle weakness first symptom, later distal hand muscle weakness
" peroneal muscles are affected much more than calf muscles
" the foot often has a typical pes cavus deformity and clawed toes
" paresthesias are uncommon, if prominent, challenge the diagnosis
" distal neuropathic pain is uncommon, but the foot deformity often causes pain when the patient walks
" essential tremor is sometimes present; previously such patients were designated as Roussy-Lévy syndrome
" usually does not affect life expectancy
" the severity of the disorder even within ecah group depends on the exact location of the mutation

**Strategy**

" demonstrate generalized demyelinating dysfunction of peripheral sensory and motor nerves. Often also autonomic nerves are affected to varying degrees.
" differentiate from acquired demyelinating polyneuropathies

**Expected abnormal findings**

*Neurography*

" sensory and motor nerve conduction velocities are reduced, usually by more than 30%
" median motor CV in most patients <38 m/s
" distal motor latencies often > 7 ms
" conduction block is uncommon
" the amplitude of the sensory nerve action potentials are reduced. Distal leg nerves are more affected than arm nerves.
" in severe demyelinating axonal polyneuropathies also motor amplitudes are reduced due to secondary axonal involvement
" F wave latencies are delayed and the number of F waves may be reduced
" A-waves maybe seen

*EMG*

" limb muscles show varying degrees of neurogenic involvement
" distal muscles are more affected than proximal muscles
" leg muscles are more affected than arm muscles

*Autonomic testing*

" may be abnormal

*Sensory thresholds*

" often abnormal

**Procedure**

*Neurography*

" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.

*EMG (optional)*

" one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

**References**

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**Modified**

- 28.3.1997 BF 3.4.97 ES, BF 7.5.1997

## HEREDITARY MOTOR AND SENSOR NEUROPATHY TYPE 2 (HMSN2, CHARCOT-MARIE-TOOTH)

**Etiology**

- genetically heterogeneous, several subtypes have been described
- most follow an autosomal dominant inheritance
- so far two types have been identified: one type is linked to chromosome 1p35-p36 and another to 3q13-22

**Clinical features**

- slowly developing sensory and motor polyneuropathy
- first symptoms usually during the second decade but may start earlier or considerably later
- peroneal muscle weakness first symptom later distal hand muscle weakness
- peroneal muscles are affected much more than calf muscles
- the foot often has a typical pes cavus deformity and clawed toes
- paresthesias are uncommon, if prominent, challenge the diagnosis
- distal neuropathic pain is uncommon, but the foot deformity causes often pain when the patient walks
- essential tremor is sometimes present, previously such patients were designated as Roussy-Lévy syndrome
- usually does not affect life expectancy

**Strategy**

- demonstrate generalized axonal dysfunction of peripheral sensory and motor nerves. Often also autonomic nerves are affected to varying degrees.
- differentiate from acquired demyelinating polyneuropathies

**Expected abnormal findings**

*Neurography*

- SCS ampl are reduced, distal leg nerves are more affected than arm nerves.
- in moderate to severe axonal polyneuropathies also motor amplitudes are reduced
- CV is slow normal or slightly reduced (not more than 30% of reference values) in axonal polyneuropathies
- median MCV >38 m/s
- F waves are delayed and the number of F waves may be reduced
- A-waves are often seen

*EMG*

- limb muscles show varying degrees of neurogenic involvement.
- distal muscles are more affected than proximal muscles.
- leg muscles are more affected than arm muscles

*Autonomic testing*

- may be abnormal

*Sensory thresholds*

- often abnormal

**Procedure**

*Neurography*

- MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
- SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.

*EMG (optional)*

- one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*

- RR-interval
- SSR
- pletysmography

*Sensory thresholds (optional)*

- temperature and vibration

**References**


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**Modified**

" 31.3.1997 ES 3.4.97

## HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 3 (HMSN3, DEJERINE-ŠOTTAS)

**Etiology**

" genetically heterogeneous
" autosomal dominant and recessive inheritance, may be sporadic
" linkage to chromosomes 17p11.2, 1q22 and 8qter have been described

**Clinical features**

" starts neonatally or in infancy
" debilitating sensory and motor polyneuropathy
" patients usually confined to wheelchair in adulthood

**Strategy**

" demonstrate generalized demyelinating dysfunction of peripheral sensory and motor nerves. Often also autonomic nerves are affected to varying degrees.
" differentiate from acquired demyelinating polyneuropathies

**Expected abnormal findings**

*Neurography*

" sensory and motor nerve conduction velocities are usually severely reduced (CV = 5-15 m/s)
" the amplitude of the sensory nerve action potentials are reduced. Distal leg nerves are more affected than arm nerves.
" in severe demyelinating axonal polyneuropathies also motor amplitudes are reduced due to secondary axonal involvement
" F wave latencies are delayed and the number of F waves may be reduced
" A-waves maybe seen

*EMG*

" limb muscles show varying degrees of neurogenic involvement.
" distal muscles are more affected than proximal muscles.
" leg muscles are more affected than arm muscles

*Autonomic testing*

" often abnormal

*Sensory thresholds*

" often abnormal

**Procedure**

*Neurography*

" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.

*EMG (optional)*

" one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

**References**

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**Modified**

" 31.3.1997 BF, 3.4.97 ES

## HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 4 (HMSN4, MB REFSUM)

**Etiology**

" phytanic acid storage disease, patients show almost no oxidation of phytanic acid
" deficiency of the peroxisomal enzyme alpha-hydroxy acid oxidase
" autosomal recessive inheritance

**Clinical features**
" onset from childhood to third decade
" pigmentary retinal degeneration, night blindness
" sensory-motor neuropathy, peripheral nerves are often palpably enlarged
" visual dysfunction precedes polyneuropathy
" cerebellar symptoms: ataxia, nystagmus and intention tremor
" pes cavus
" often ichtyosis of the skin, anosmia, cardiomyopathy and sensorineural hearing loss

**Strategy**
" demonstrate demyelinating polyneuropathy
" demonstrate pigmentary retinopathy

**Expected abnormal findings**
*Neurography*
" sensory and motor nerve conduction velocities are moderately reduced, usually by more than 30%
" the amplitude of the sensory nerve action potentials are reduced. Distal leg nerves are more affected than arm nerves.
" in severe demyelinating axonal polyneuropathies also motor amplitudes are reduced due to secondary axonal involvement
" F wave latencies are prolonged and the number of F waves may be reduced
" A-waves maybe seen
*EMG*
" limb muscles show varying degrees of neurogenic involvement. distal muscles are more affected than proximal muscles
" leg muscles are more affected than arm muscles
*Electroretinography (ERG)*
" absence of both cone and rod responses
*BAEP (brainstem auditory evoked potentials)*
" abnormal
*Autonomic testing*
" often abnormal
*Sensory thresholds*
" often abnormal

**Procedure**
*Neurography*
" MCS: n.medianus, n.ulnaris, n.tibialis, n.peroneus unilaterally
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.
*EMG (optional)*
" one distal and proximal muscle in the lower extremities.
*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography
*Sensory thresholds (optional)*
" temperature and vibration
*Electroretinography (ERG)*
" absence of both cone and rod responses

**References**
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**Modified**
" 2.4.1997, ES 3.4.97

## HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES (HNPP)

**Etiology**
" autosomal dominant inheritance, linked to chromosome 17p11.2
" gene product peripheral myelin protein 22 (PNP-22)
" locus of deletion is identical to HMSN 1a which has a duplication at the same site

**Clinical features**
" liability to develop local nerve lesions following compression or trauma that usually would not cause a nerve lesion
" presents often in the late teens or young adult age, sometimes seen also in young children
" some patients may have several nerve lesions
" usually the recovery of the local nerve lesions is good
" many carriers of the gene do not have any local nerve lesions
" before the age of 30 neurological examination is normal except for the local nerve lesions
" in old age there are distinct clinical signs of polyneuropathy, decreased tendon reflexes and peroneal muscle weakness

**Strategy**
" demonstrate a mild generalized demyelinating dysfunction of peripheral sensory and motor nerves
" most clearcut findings will be found at common entrapment sites (n.medianus at the wrist, n.ulnaris at the elbow)
" most patients will present with mononeuropathies, most commonly radial nerve lesions, peroneal lesions, median nerve lesions
" differentiate from acquired demyelinating polyneuropathies

**Differential diagnosis**
" other polyneuropathies
" HMSN1, usually has more slowing of nerve conduction
" hereditary recurrent

**Expected abnormal findings**

*Neurography*

" sensory and motor nerve conduction velocities are reduced to varying degrees
" some nerves may have CVs in the low normal range
" n.radialis and suralis SCV is usually reduced by 20-30%
" the conduction velocities at common entrapment sites are often considerably reduced
" median nerve motor distal latencies can be prolonged up to 5-7 ms without symptoms of CTS
" the amplitude of the sensory nerve action potentials are reduced in middle-aged and older subjects

*EMG*

" findings depend on the type of local nerve lesions
" middle-aged and older patients show signs of neurogenic involvement in distal leg muscles due to the polyneuropathy (sometimes it may difficult to tell whether the findings in the leg muscles are due to mild repeated peroneal nerve lesions or polyneuropathy)

*Sensory thresholds*

" vibration thresholds and thermal thresholds are normal in young subjects but show abnormalities in middle-aged or older subjects

**Procedure**

*Neurography*

" MCS bilateral: n.medianus, n.ulnaris (fractionated across the elbow) and peroneal nerves (fractionated across the fibular head),
" SCS: n.medianus and n.ulnaris bilaterally and n.radialis, n.suralis unilaterally.

*EMG (optional)*

" one distal and proximal muscle in the lower extremities.

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

*Sensory thresholds (optional)*

" temperature and vibration

**References**


" Chance PF, Alderson MK, Leppig KA, et al. DNA deletion associated with hereditary neuropathy with liability to pressure palsies. Cell 1993;72:143-151
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**Modified**

" 31.3.1997 BF, 3.4.97 ES

## INHERITED RECURRENT BRACHIAL PLEXUS NEUROPATHY

**Etiology**

" autosomal dominant inheritance
" linked to chromosome 17q24

**Clinical features**

" acute, usually painful episodes of local neuropathies
" most nerve lesions are confined to plexus brachialis and the upper extremities, but any nerve may be affected
" a distinctive feature is that nerve lesions appear often during pregnancy and puerperium
" infections, surgery, immunization and strenuous exercise may trigger episodes
" the episodes resemble sporadic episodes of “neuralgic amyotrophy”
" onset varies considerably, often first episode in early childhood
" prognosis of individual nerve lesions is generally good
" many reports describe mild facial dysmorphic features, in our experience they are not essential

**Strategy**

" demonstrate local or regional neuropathy
" exclude generalized polyneuropathy

**Expected abnormal findings**

*Neurography*

" depends on the nerve affected

*EMG*

" muscles chosen in relation to the affected nerve or plexus structure

**Expected normal findings**

*EMG*

" normal outside affected nerves

*Neurography*

" normal outside affected nerves

*Autonomic tests*

" RR-interval
" SSR
" pletysmography

**Procedure**

*Neurography*

" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.

*EMG*

" one distal and proximal muscle in the lower extremities.

*Sensory thresholds (optional)*

" temperature and vibration

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

**References**


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**Modified**

" 31.3.1997 BF, 3.4. 97 ES

## CRITICAL ILLNESS POLYNEUROPATHY

**Etiology**

" exact mechanism of neuropathy is not clear
" occur during intensive care in association with sepsis and multiple organ failure
" axonal polyneuropathy

**Clinical features**

" neuropathy becomes apparent after the sepsis and multiple organ failure has been controlled
" often the neuropathy is recognized when attempts to wean the patient from the respirator are made
" the patient is awake and often unable to maintain spontaneous breathing

**Strategy**

" demonstrate acute sensory and motor axonal polyneuropathy

**Expected abnormal findings**

*Neurography*

" SCS: AMPL reduced or missing responses
" SCS: CV may be reduced
" MCS: low amplitudes
" MCS: slight CV reduction may be present
" F waves: reduced number of F waves, related to low AMPL
" Autonomic test usually show abnormalities

**Expected normal findings**

" none

**Procedure**

*Neurography*

" MCS: n.medianus, n.ulnaris and n.peroneus and n.tibialis unilaterally
" SCS: n.radialis and n.suralis bilaterally.

*EMG*

" one distal and proximal muscle in the lower extremities
" one distal and proximal muscle in the upper extremities

*Sensory thresholds (optional)*

" temperature and vibration

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

**References**


" Bolton CF, Gilbert IJ, Hahn AF, Sibbaldi WJ. Polyneuropathy in critcally ill Patients. J Neurol Neurosurg Psychiatry 1984; 47: 1223-1231
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**Modified**

" 2.4.1997, 3.4.97 ES

## SENSORY AXONAL POLYNEUROPATHY

**Etiology**
" paraneoplastic, often related to small cell lung cancer
" Sjögrens's syndrome

**Clinical features**
" loss of sensory modalities with a distal distribution
" paresthesias

**Strategy**
" demonstrate generalized dysfunction of peripheral sensory nerves.
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**
*Neurography*
" SCS: AMPL reduced
" SCS: CV may be reduced
*Sensory thresholds*
" vibration thresholds and thermal thresholds are abnormal

**Expected normal findings**
*EMG*
*Neurography*
" MVC
*Autonomic tests*
" RR-interval
" SSR
" pletysmography

**Procedure**
*Neurography*
" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.
*EMG*
" one distal and proximal muscle in the lower extremities.
*Sensory thresholds (optional)*
" temperature and vibration
*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

**References**
" Chalk CH, Windebank AJ, Kimmel DW, McManis PG. The distinctive clinical features of paraneoplastic sensory neuronopathy. Can J Neurol Sci 1992; 19:346-351
" Chalk CK, Lennon VA, Stewens JC, Windebank AJ. Seronegative for type 1 antineuronal nuclear antibodies (anti-Hu) in subacute sensory neuronopathy patients without cancer. Neurology 1993;43:2209-2211
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**Modified**
" 2.4.1997, 3.4.97 ES

## HEREDITARY SENSORY POLYNEUROPATHY TYPE 1 (HEREDITARY SENSORY NEUROPATHY OF DENNY-BROWN)

**Etiology**
" autosomal dominant inheritance
" mapped to chromosome 9q22.1-q22.3 in one family

**Clinical features**
" onset during second decade
" progressive distal extremity sensory loss
" pain and temperature more affected than touch and pressure
" mutilation of feet
" autonomic function usually preserved except sweating distally

**Strategy**
" demonstrate generalized dysfunction of peripheral sensory nerves
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**
*Neurography*
" the amplitude of the sensory nerve action potentials are reduced in young patients
" no sensory nerve action potentials obtainable in older patients
*Sensory thresholds*
" vibration thresholds and thermal thresholds are abnormal

**Expected normal findings**
*Neurography*
" MVC
*EMG*
" all muscles
*Autonomic tests*
" RR-interval
" SSR
" pletysmography

**Procedure**

*Neurography*
" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.
*EMG*
" one distal and proximal muscle in the lower extremities.
*Sensory thresholds (optional)*
" temperature and vibration
*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

**References**

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**Modified**
" 1.4.1997 BF, 3.4.97 ES

## HEREDITARY SENSORY POLYNEUROPATHY TYPE 2

**Etiology**
" autosomal recessive inheritance

**Clinical features**
" rare
" onset in childhood or at birth
" all sensory modalities are affected, touch and pressure earlier than pain and temperature
" hands and feet mutilated

**Strategy**
" demonstrate generalized dysfunction of peripheral sensory nerves
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**
*Neurography*
" unobtainable sensory nerve action potentials
*Sensory thresholds*
" vibration thresholds and thermal thresholds are abnormal

**Expected normal findings**
*Neurography*
" MVC
*EMG*
*Autonomic tests*
" RR-interval
" SSR
" pletysmography

**Procedure**
*Neurography*
" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.
*EMG*
" one distal and proximal muscle in the lower extremities.
*Sensory thresholds (optional)*
" temperature and vibration
*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

**References**

" Bye AM, De C Baker W, Pollard J, Wise G. Hereditary sensory neuropathy type II, without trophic changes. Dev Med Child Neurol 1990;32:164-171
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**Modified**
" 1.4.1997 BF, 3.4.97 ES

## HEREDITARY SENSORY POLYNEUROPATHY TYPE 3 (FAMILIAL DYSAUTONOMIA, RILEY-DAY SYNDROME)

**Etiology**
" autosomal recessive inheritance, majority of patients are Jewish
" linked to chromosome 9q31-33
" very rare

**Clinical symptoms**
" symptoms present from birth

" autonomic dysfunction very prominent
" absent lacrimation, corneal ulceration
" labile sweating, blood pressure and temperature
" diminution of pain and temperature sensation
" touch and pressure preserved
" mutilation unusual
" intelligence normal
" scoliosis common
" decreased life expectancy

**Strategy**

" demonstrate generalized dysfunction of peripheral sensory nerves
" demonstrate generalized dysfunction of the autonomic nervous system
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**

*Neurography*

" the amplitude of the sensory nerve action potentials are reduced

*Sensory thresholds*

" vibration thresholds and thermal thresholds are abnormal

*Autonomic function*

" RR-interval
" SSR
" pletysmography

**Expected normal findings**

*Neurography*

" MVC

*EMG*

" all muscles

**Procedure**

*Neurography*

" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.

*EMG*

" one distal and proximal muscle in the lower extremities.

*Sensory thresholds (optional)*

" temperature and vibration

*Autonomic tests (optional)*

" RR-interval
" SSR
" pletysmography

**Note**

" A prompt miosis of the pupil in response to 2.5% metacholine is a characteristic

**References**


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**Modified**

" 1.4.1997, 3.4.97 ES

## HEREDITARY SENSORY POLYNEUROPATHY TYPE 4 (CONGENITAL SENSORY NEUROPATHY WITH ANHIDROSIS)

**Etiology**

" autosomal recessive inheritance

**Clinical features**

" symptoms present from birth
" widespread absence of pain and temperature sensation
" preserved touch and pressure sensation
" episodic fever
" self mutilation common
" mental retardation
" short stature

**Strategy**

" demonstrate generalized dysfunction of the autonomic nervous system
" demonstrate dysfunction of unmyelinated and thin myelinated sensory axons
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**

*Sensory thresholds*

" thermal thresholds are abnormal

*Autonomic function*

" RR-interval

" SSR
" pletysmography

**Expected normal findings**

*Neurography*
" MVC
" SCV

*EMG*
" all muscles

*Sensory thresholds*
" vibration thresholds

**Procedure**

*Neurography*
" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.

*EMG*
" one distal and proximal muscle in the lower extremities.

*Sensory thresholds (optional)*
" temperature and vibration

*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

**References**

" Indo Y, Tsuruta M, Hayshida Y, et al. Mutations in the TRKA/NGF receptor gene in patients with congenital insensitivity to pain with anhidrosis. Nature Genetics 1996;13:485-488
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**Modified**

" 1.4.1997 BF, 3.4.97 ES

## TANGIER DISEASE

**Etiology**

" autosomal recessive inheritance
" hereditary high-density lipoprotein deficiency and widespread tissue storage of cholesteryl esters

**Clinical features**

" onset is mostly after the age of 20 years
" progressive weakness of proximal limb muscles
" pseudo-syringomyelic picture with a dissociated sensory loss

**Strategy**

" demonstrate evidence of an axonal neuropathy
" differentiate from sensory-motor axonal polyneuropathies

**Expected abnormal findings**

*Neurography*
" MVC: normal or slight abnormalities compatible with axonal neuropathy
" SCV: reduced amplitude, normal or slightly reduced CV

*EMG*
" Findings compatible with chronic neuropathy

*Sensory thresholdsvibration thresholds*
" thermal thresholds are abnormal

*Autonomic function*
" RR-interval
" SSR
" pletysmography

**Expected normal findings**

*Neurography*
" sometimes normal MCS

*EMG*
" sometimes normal findings

**Procedure**

*Neurography*
" MCS bilateral: n.medianus, n.ulnaris and n.peroneus
" SCS: n.medianus, n.ulnaris. n.radialis and n.suralis bilaterally.

*EMG*
" one distal and proximal muscle in the lower extremities.

*Sensory thresholds (optional)*
" temperature and vibration

*Autonomic tests (optional)*
" RR-interval
" SSR
" pletysmography

**References**

" Fazio R, Nemni R, Quattrini A, et al. Acute presentation of Tangier polyneuropathy: a clinical and morphological study. Acta Neuropathol 1993;86:90-94
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**Modified**
" 1.4.1997 BF, 3.4.97 ES

## DIPHTHERIA

**Etiology**
" exotoxin produced by Corynebacterium diphteriae
" the exact mechanism of peripheral neuropathy is uncertain

**Clinical features**
" incubation period 2-6 days
" fever sore throat and characteristic membranous pharyngitis
" 2-4 weeks following the initial infection paralysis occurs in 10-20 % of patients
" major attack is on cranial and peripheral nerves
" most frequent manifestation is palatal paralysis,
" extraocular, pharyngeal and diaphragmatic muscles may be involved
" sometimes peripheral neuropathy resembling polyradiculitis develops 3-8 weeks after the infection
" palatal weakness is the only weakness before 2 weeks after onset
" during 3 to 5 weeks there is pharyngeal paresthesia and paralysis of extraocular muscles,
" during 5-7 weeks weakness of the larynx and diaphragm occur
" sensory-motor polyneuropathy develops 2-3 months after the onset

**Strategy**
" demonstrate sensory-motor polyneuropathy
" differentiate from polyradiculitis

**Expected abnormal findings**
*EMG*
" neurogenic EMG findings in muscles
*Neurography*
" SCS: reduced CV and ampl
" MCS: reduced CV and amplitude

**Procedure**
*EMG*
" m.deltoideus
" m.interosseus
" m.vastus lateralis
" m.tibialis anterior
*Neurography*
" SCS: n.suralis, n.radialis bilaterally
" MCS: n.medianus and n.peroneus bilaterally

**References**
" Kurdi A, Abdul-Kader A. Clinical and electrophysiological studies in diphtheria neuritis in Jordan. J Neurol Sci 1979;42: 243-250
" Swift TR, Rivner MH. Infectious disease of nerve. 179-194 in Vinken PJ, Bruyn GW, Klawans HL (Edittors) Handbook of Clinical Neurology revised series 7. Part 51 Neuropathies (Edited by Matthews WB).er Science Publishers, Amsterdam, 1987.

**Modified**
" 2.4.1997, 3.4.97 ES

## LEPROSY (HANSEN'S DISEASE)

**Etiology**
" infection by Mycobacterium leprae
" although rarely seen in industrialized countires, it is most common neuropathy in the world
" trnasmiited through nasal secretions and cutaneous contact
" only a small minority of exposed subjects will be infected
" manifestations depend on the immunological reaction of the host

**Clinical features**
*Lepromatous leprosy*
" low host immune reacton, abundant bacilli
" skin in coooler areas (fingers, toes, pinnae of the ears and nose) of the body become infiltrated with bacilli
" damage to to unmyelinated nerves initially
" characteristically loss of pain and temperature sensation
" loss of pain sensation results in mutilation
" peripheral nerves are thickend
" ulnar nerve at the wrist and elbow and n.peroneus at the knee mey be affected leading to motor deficits
*Tuberculoid leprosy*
" strong cell mediated immune reaction leading to localized reaction with tissue damage
" cutaneous depigmented lesions that are anhidrotic and anestetic
" distribution of skin changes is asymmetric
" most often affected nerves are n.facialis, n.medianus, n.ulnaris and n.peroneus
*Borderline leprosy*
" intermediate form between these two

**Strategy**
" demonstrate affection of distal sensory nerves
" variable affection of n.ulnaris, n.medianus n.peroneus, motor axons may be affected

**Expected abnormal findings**

*EMG*

" distal hand muscles and the peroneal muscles if the nerves are locally affected

*Neurography*

" SCS in the distal parts of the limbs, especially the digital nerves

**Procedure**

*EMG*

" Depends on the region affected

*Neurography*

" SCS: n.suralis, n.radialis, n.peroneus superficialis, n.medianus, n.ulnaris bilaterally
" MCS: n.medianus, n.ulnaris and n.peroneus bilaterally

**References**


" Ridley DS, Jopling WH: Classification of leprosy according to immunity: A five-group system. Int J Lepr Other Mycobact Dis 1966;34:255-258
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**Modified**

" 15.4.1997

# 3. NEUROMUSCULAR TRANSMISSION DISORDERS

## MYASTHENIA GRAVIS (MG)

**Etiology**

" humoral autoimmune response against acetylcholine receptors

**Clinical features**

" incidence 2-10/1000000 per year
" prevalence 25-125/1000000
" female preponderance, female to male ratio is 6:4
" female incidence peaks around 30 years, male around 60-70 years
" abnormal fatigability and weakness of some or all voluntary muscles
" muscles innervated by cranial nerves and proximal muscles are more affected than distal muscles
" bilateral or unilateral ptosis and ocular palsies are often the initial symptom
" weakness increases during repeated or sustained exertion
" symptoms are aggravated by heat and often improved in cold
" spontaneous remissions may occur for varying periods, complete remissions are rare

**Strategy**

" demonstrate postsynaptic neuro-muscular transmission defect
" exclude other causes of fatigue

**Expected abnormal findings**

*Repetitive stimulation*

" abnormal decrement m.deltoideus/m.nasalis/m.trapezius/m.anconeus (abnormal in 65-80% of patients with generalized MG)

*SFEMG*

" increased jitter

*EMG*

" increased “jiggle”
" a few fibrillations and positive sharp waves may be seen, rarely moderate amounts
" MUPs may be brief

*Neurography*

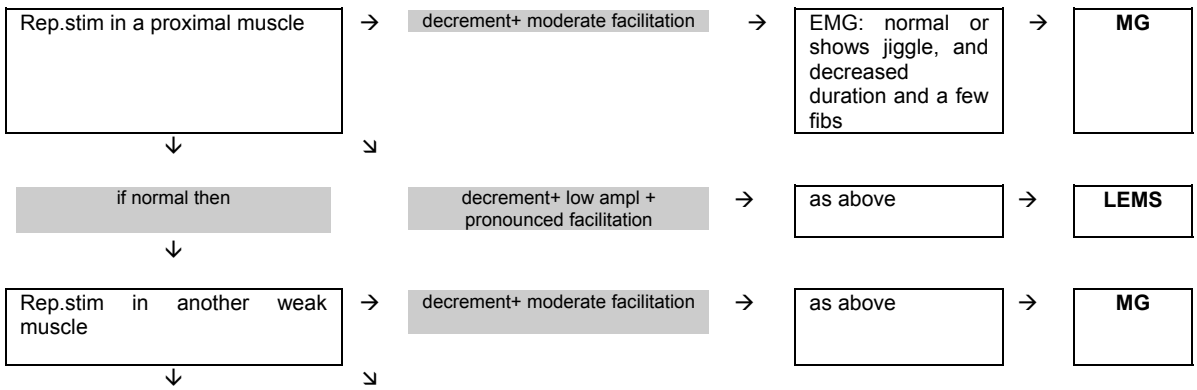
" MCS ampl may be reduced in severe MG

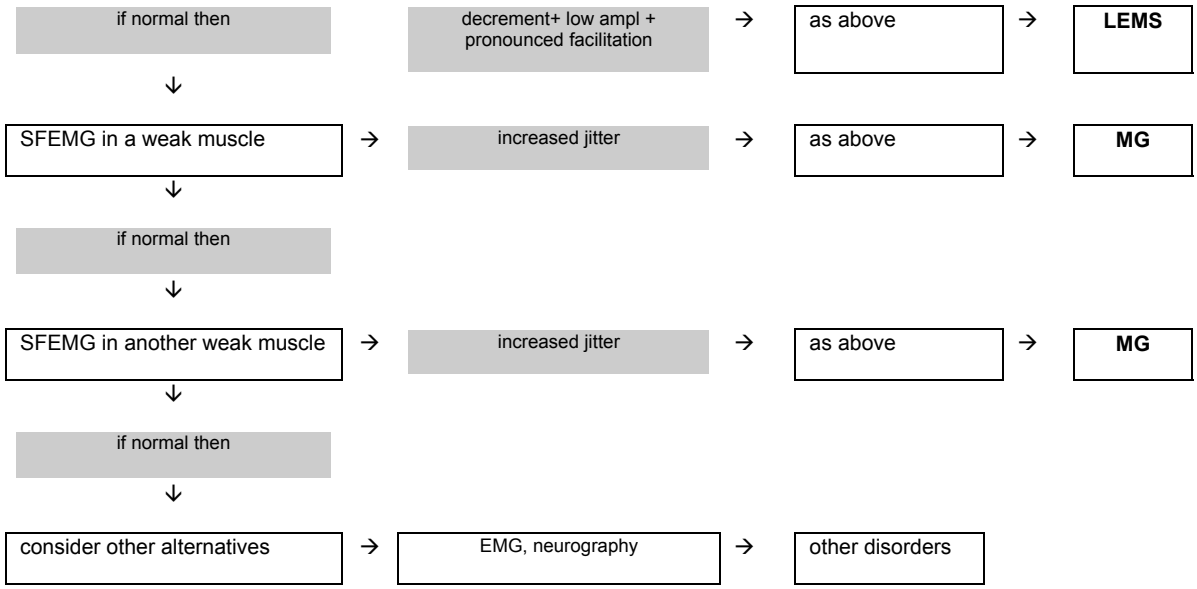
**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**






**Note**

" distal muscles show decrement only in 15-20 % of patients with generalized MG

**References**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**Modified**

" 2.4.1997 BF, 3.4.97 ES

## NEONATAL MYASTHENIA GRAVIS

**Etiology**

" occurs in neonates whose mother have myasthenia gravis
" antibodies against acetylcholine receptors diffuse across the placenta from the mother
" neonatal myasthenia develops in 10-15 % of children born to mothers with myasthenia gravis

**Clinical features**

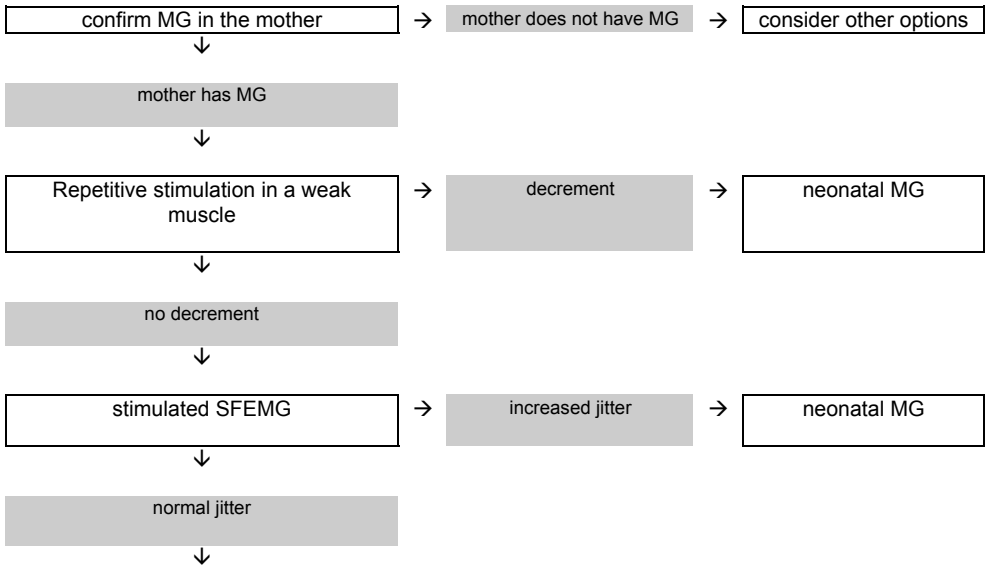
" symptoms appear within the first hours after birth
" generalized weakness, feeding and respiratory weakness
" lasts usually for 3-4 weeks

**Strategy**
" suspect disorder if the mother has myasthenia gravis
" demonstrate postsynaptic neuro-muscular transmission defect

**Expected abnormal findings**
*Repetitive stimulation*
" increased decrement in m.deltoideus/m.nasalis/m.trapezius/m.anconeus
*SFEMG*
" increased jitter
*EMG*
" increased "jiggle"
" MUPs may be brief

**Expected normal findings**
*Neurography*
" SCS
" MCS

**Procedure**




**References**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" Bransch CE Jr; Swift TR; Dyken PR Prolonged neonatal myasthenia gravis: electrophysiological studies. Ann Neurol 1978;3:416-418
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**Modified**
" 2.4.1997 BF

## MYASTHENIC SYNDROME ( LAMBERT-EATON MYASTHENIC SYNDROME, LEMS)

**Etiology**
" humoral autoimmune response against presynaptic calcium channels
" disturbed neuromuscular transmission

**Clinical features**
" rare
" male preponderance male to female ratio 4,7:1
" 70 % of males and 40 % of females have an associated malignancy, small cell carcinoma of the lung is most common
" weakness and fatiguability of limb and truncal muscles
" symptoms due to dysfunction of the autonomic nervous system are common: reduced lacrimation, dryness of the mouth, impotence and orthostatism

**Strategy**
" demonstrate presynaptic neuro-muscular transmission defect
" exclude other causes of fatigue

**Expected abnormal findings**
*Repetitive stimulation*
" increased decrement (even distal muscles are often affected in LEMS)
" abnormal facilitation following exercise
" small CMAP amplitude

*SFEMG*
" increased jitter
*EMG*
" increased "jiggle"
" a few fibrillations and positive sharp waves may be seen, rarely moderate amounts
" MUPs may be brief
*Neurography*
" MCS amplitude is often reduced

**Expected normal findings**

*Neurography*
" SCS
" MCS, except amplitude

**Procedure**

| | | | | | | |
|---|---|---|---|---|---|---|
| Rep.stim in a distal muscle | → | decrement+ moderate facilitation | → | EMG: normal or shows jiggle, and decreased duration and a few fibs | → | **MG** |
| ↓ ↘ | | | | | | |
| if normal then | | decrement+ low ampl + pronounced facilitation | → | as above | → | **LEMS** |
| ↓ | | | | | | |
| Rep.stim in another weak muscle | → | decrement+ moderate facilitation | → | as above | → | **MG** |
| ↓ ↘ | | | | | | |
| if normal then | | decrement+ low ampl + pronounced facilitation | → | as above | → | **LEMS** |
| ↓ | | | | | | |
| SFEMG in a weak muscle | | increased jitter | → | as above | → | **MG** |
| ↓ | | | | | | |
| if normal then | | | | | | |
| ↓ | | | | | | |
| SFEMG in another weak muscle | → | increased jitter | → | as above | → | **MG** |
| ↓ | | | | | | |
| if normal then | | | | | | |
| ↓ | | | | | | |
| Consider other alternatives | → | EMG, neurography | → | other disorders | | |

**Note**

" in contrast to MG, LEMS shows decrement also in distal muscles

**References**


" Simpson JA. Electrophysiology of neuromuscular junction disease: an appreciation of the contributions of Edward H. Lambert. Muscle Nerve 1982;5:S6-11Schwartz MS; Stålberg E. Myasthenic syndrome studied with single fiber electromyography. Arch Neurol 1975;32:815-817
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" Howard JF Jr; Sanders DB; Massey JM. The electrodiagnosis of myasthenia gravis and the Lambert-Eaton myasthenic syndrome. Neurol Clin 1994;12:305-330
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**Modified**

" 2.4.1997 BF, 3.4.97

## SLOW CHANNEL SYNDROME

**Etiology**
- autosomal dominant inheritance with complete penetrance
- variable expression, sporadic cases do occur
- MEPP and EPP durations are prolonged
- MEPP amplitude decreased in severely affected muscles
- quantal content normal
- prolonged open time of AchR ion channels probably the cause of abnormalities *(slow channels)*
- on electron microscopy junctional folds are abnormal, the number acetylcholine receptors is reduced

**Clinical features**
- onset may be from infancy to adulthood
- severity and progression varies considerably
- severe weakness and fatiguability of cervical, scapular and finger extensor muscles
- the fatiguability and weakness fluctuates but much less than in myasthenia gravis
- mild involvement of eyelid and extraocular muscles
- leg muscles are less affected
- reduced tendon reflexes
- acetylcholinesterase inhibitors have no effect

**Strategy**
- demonstrate neuromuscular transmission defect
- *demonstrate at single stimuli repetitive motor responses*

**Expected abnormal findings**

*EMG*
- unstable MUP shape in weak muscles

*SF-EMG*
- increased jitter

*Repetitive stimulation*
- abnormal decrement at 2-3 Hz stimulation
- postactivation exhaustion initially improves decrement then within a minute increased decrement
- *at single stimuli there are repetitive motor responses ( seen as abnormal shape of the CMAP) that disappear at stimulus frequencies above 0,2 Hz*

**Expected normal findings**

*Neurography*
- SCS
- MCS

**Procedure**

*EMG*
- m.deltoideus/m.biceps/m.trapezius
- m.interosseus/m.extensor digitorum communis
- m.vastus lateralis
- m.tibialis anterior

*Neurography*
- SCS: n.suralis, n.radialis unilaterally
- MCS: n.medianus and n.peroneus unilaterally

*Repetitive stimulation*
- m.trapezius/m.deltoideus
- thenar muscles
- check for repetitive responses at low stimulus frequencies (present in slow channel syndrome and congenital AChE deficiency)

**Note**
- M-response shape may be abnormal with irregular terminal part due to extra discharges.

**References**

- Engel AG, Lambert EH, Mulder DM, et al. A newly recognized congenital myasthenic syndrome attributed to a prolonged open time of acetylcoline induced ion channel. Ann Neurol 1982,11:553-569
- Gomez CM, Maselli R, Gammack JT, et al. A beta-subunit mutation in the acetylcholine receptor channel gate causes severe slow-channel syndrome. Ann Neurol 1996,39:712-723
- Misulis KE, Fenichel GM. Genetic forms of myasthenia gravis. Pediatr Neurol 1989,5:205-10
- Shillito P, Vincent A, Newsom-Davis J. Congenital myasthenic syndromes. Neuromusc Disord 1993,3:183-90


**Modified**
- 30.3.1997 BF, 3.4.97 ES

## CONGENITAL ACETYCHOLINERECEPTOR (ACR) DEFICIENCY

**Etiology**
- probably autosomal recessive inheritance, but it is more common in men than women
- number of AcCR reduced
- small MEPPs
- poorly developed junctional folds

**Clinical features**
- onset at birth or before 2 years
- ptosis and bulbar muscle involvement
- mild to moderate fatiguable weakness
- generally benign course, persists into adult life
- no atrophy or myopathy
- acetylcholine esterase inhibitors improve symptoms

**Strategy**

" demonstrate neuromuscular transmission defect

**Expected abnormal findings**

*EMG*

" unstable MUP shape in weak muscles

*SF-EMG*

" increased jitter

*Repetitive stimulation*

" abnormal decrement at 2-3 Hz stimulation

**Expected normal findings**

*Neurography*

" SCS

" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius

" m.interosseus/m.extensor digitorum communis

" m.vastus lateralis

" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally

" MCS: n.medianus and n.peroneus unilaterally

*Repetitive stimulation*

" m.trapezius/m.deltoideus

" thenar muscles

" check for repetitive responses at low stimulus frequencies (present in slow channel syndrome and congenital AChE deficiency)

**References**

" Lecky BRF, Morgan-Hughes JA, Murray NMF. Congenital myasthenia: Further evidence of disease heterogeneity. Muscle Nerve 1986;9:233-242

" Shillito P, Vincent A, Newsom-Davis J. Congenital myasthenic syndromes. Neuromusc Disord 1993;3:183-90

" Vajsar J, Sloane A, MacGregor DL, Ronen GM, et al. Arthrogryposis multiplex congenita due to congenital myasthenic syndrome. Pediatr Neurol 1995;12:237-241

" Vincent A, Cull-Candy SG, Newsom-Davis J, et al. Congenital myasthenia: End-plate acetyl-choline receptors and electrophysiology in five cases. Muscle Nerve 1981;4:306-318

**Modified**

" 30.3.1997 BF, 3.4.97

## FAMILIAL INFANTILE MYASTHENIA

**Etiology**

" autosomal recessive inheritance

" miniature end-plate potential amplitude decreases during prolonged activity

" probably progressive decrease of acetylcholine content in synaptic vessels during prolonged stimulation (defect in acetylcholine re-uptake or synthesis)

**Clinical features**

" presents in infancy or early childhood

" fluctuating ptosis

" involvement of bulbar muscles, weakness of sucking and breathing

" course worsened often by infections and fever

" apneas from respiratory muscle weakness may cause sudden death

" during childhood patients appear normal between crises or have mild weakness

" after 10 years of age patients have easy fatiguability

" responds to acetylcholinesterase inhibitors

" normal tendon reflexes, no muscle atrophy

**Strategy**

" demonstrate neuromuscular transmission defect

**Expected abnormal findings**

*EMG*

" unstable MUP shape in weak muscles

*SF-EMG*

" increased jitter

*Repetitive stimulation*

" abnormal decrement at 2-3 Hz stimulation

" postactivation exhaustion initially improves decrement then within a minute increased decrement

**Expected normal findings**

*Neurography*

" SCS

" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius

" m.interosseus/m.extensor digitorum communis

" m.vastus lateralis

" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally

" MCS: n.medianus and n.peroneus unilaterally

*Repetitive stimulation*

" m.trapezius/m.deltoideus
" thenar muscles
" check for repetitive responses at low stimulus frequencies (present in slow channel syndrome and congenital AChE deficiency)

**References**

" Chan-Lui WY, Hawkins BA. Infantile myasthenia. Neuropediatrics 1985;16:24-28
" Fenichel GM. Clinical syndromes of myasthenia in infancy and childhood. Arch Neurol 1978;35:97-103
" Misulis KE, Fenichel GM. Genetic forms of myasthenia gravis. Pediatr Neurol 1989;5:205-10
" Zammarchi E, Donati MA, Masi S, Sarti A, Castelli S. Familial infantile myasthenia: a neuromuscular cause of respiratory failure. Child's Nerv Syst 1994;10:347-9

**Modified**

" 30.3.1997 BF, 3.4.97

## CONGENITAL ENDPLATE ACETYLCHOLINE ESTERASE DEFICIENCY (LIMB-GIRDLE MYASTHENIA)

**Etiology**

" autosomal recessive inheritance
" lack of acetylcholine esterase at the neuromuscular junctions
" MEPP has a normal amplitude but decay the phase is prolonged
" EPP has a reduced amplitude because the quantal content is decreased

**Clinical features**

" onset from birth or before two years
" selective involvement of axial muscles leading to scoliosis in older patients
" bulbar muscles affected
" motor milestones delayed
" the symptoms remain relatively static until the end of the first decade after that progression of symptoms
" acetylcholinesterase inhibitors do not improve symptoms, some patients may get worse
" no muscle atrophy

**Strategy**

" demonstrate neuromuscular transmission defect

**Expected abnormal findings**

*EMG*

" unstable MUP shape in weak muscles

*SF-EMG*

" increased jitter

*Repetitive stimulation*

" abnormal decrement at 2-3 Hz stimulation
" postactivation exhaustion initially improves decrement then within a minute increased decrement
" *at single stimuli there are repetitive motor responses that disappear at stimulus frequencies above 0,2 Hz*

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

*Repetitive stimulation*

" m.trapezius/m.deltoideus
" thenar muscles
" check for repetitive responses at low stimulus frequencies (present in slow channel syndrome and congenital AChE deficiency)

**Note**

" M-response shape may be abnormal with irregular terminal part due to extra discharges

**References**

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**Modified**

" 30.3.1997, 3.4.97 ES

## BOTULINUM INTOXICATION

**Etiology**

" seven different types neurotoxins A-G. produced by Clostridium botulinum
" the toxin is taken up by peripheral cholinergic nerve terminals where it initially blocks acetylcholine release and subsequently causes reversible denervation of muscle fibers

**Clinical features**

" infection is acquired when improperly processed foods are eaten
" wound botulism has been described
" in infants the spores of Clostridium botulinum may colonize the gut
" the toxin is destroyed by heat
" after an symptomatic 12-36 hour (extremes 2 hours to 2 weeks) incubation period nonspecific symptoms of nausea, diarrhea
" opthalmoplegia, bulbar palsy, respiratory paralysis, paralysis of limb muscles
" pupils dilated
" autonomic symptoms, bradychardia, hypotension, hyperhidrosis
" sensation normal

**Strategy**

" demonstrate neuromuscular transmission defect and neurogenic EMG findings in muscles

**Expected abnormal findings**

*EMG*

" fibrillations and positive sharp waves 7-10 days following the onset
" small polyphasic MUPs
" jiggle

*Neurography*

" MCS: reduced amplitude

*Repetitive stimulation*

" decrement at 2-3 Hz
" facilitation after short voluntary activation or at high stimulation frequencies

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

*Repetitive stimulation*

" m.trapezius/m.deltoideus
" thenar muscles

**References**

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**Modified**

" 2.4.1997, 3.4.97 ES

# 4 MYOPATHIES

## 4.1 INFLAMMATORY MYOPATHIES

### POLYMYOSITIS

**Etiology**
" cell mediated autoimmune response against muscle fibers

**Clinical features**
" incidence 1-9/1000000
" more common in women than men
" usually begins after the age of 20 years
" muscle weakness develops subacutely or insidiously
" proximal limb and neck flexor muscles involved more than other muscles
" muscle pain is sometimes present, but it is not as prominent as one would expect
" dysphagia is common in polymyositis
" CK is usually moderately elevated

**Strategy**
" demonstrate myopathy
" assess: severity (mild, moderate, severe) and activity (stationary, active)

**Expected abnormal findings**
*EMG*
" fibrillations in the acute stage, they disappear in remission
" small, brief MUPs in the acute stage
" later long duration polyphasic MUPs in the chronic stage
*Neurography*
" if distal muscles are severely involved MCS ampl are reduced

**Expected normal findings**
*Neurography*
" SCS
" MCS

**Procedure**
*EMG*
" m.vastus lateralis/m.vastus medialis
" m.deltoideus/m.biceps
" m.interosseus dorsalis I/m.abductor digiti minimi
" m.tibialis anterior
" paravertebral muscles in the low thoracic region should be studied if limb muscles do not show distinct abnormalities
*Neurography*
" MCS: n.medianus and n.peroneus unilaterally
" SCS: n.radialis and n.suralis unilaterally

**NOTE**
" in chronic PM, there may be a mixed pattern of small and large MUPs
" it is not unusual for patients with PM to have concurrent PNP, especially if they have malabsorption

**References**

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**Modified**
" 1.4.1997, 3.4.97 ES

## DERMATOMYOSITIS

### Etiology
" humoral mediated autoimmune response against arterioles in muscles and skin

### Clinical features
" misery
" muscle weakness: limb, bulbar and respiratory develops subacutely or insidiously
" proximal limb and neck flexor muscles involved more than other muscles
" muscle pain and tenderness are sometimes present
" arthralgia
" skin rash, typically around the eyelids (violaceous)
" skin rash over joints of fingers, knees and ankles
" vasculitis/skin ulcerations
" later in the disease calcinocis of the skin
" CK is usually moderately elevated
" course of the disease is variable

### Strategy
" demonstrate myopathy with spontaneous activity
" assess: severity (mild, moderate, severe) and activity (stationary, active)

### Expected abnormal findings
*EMG*
" fibrillations in the acute stage, these disappear in remission
" small, brief MUPs in the acute stage
" later long duration, polyphasic MUPs in the chronic stage

*Neurography*
" if distal muscles are severely involved MCS ampl are reduced

### Expected normal findings
*Neurography*
" SCS
" MCS

### Procedure
*EMG*
" m.vastus lateralis/m.vastus medialis
" m.deltoideus/m.biceps
" m.interosseus dorsalis I/m.abductor digiti minimi
" m.tibialis anterior
" paravertebral muscles in the low thoracic region

*Neurography*
" MCS: n.medianus and n.peroneus unilaterally
" SCS: n.radialis and n.suralis unilaterally

### References

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### Modified
" 2.4.1997, 3.4.97 ES

## INCLUSION BODY MYOSITIS AND INCLUSION BODY MYOPATHIES (IBM)

### Etiology
" IBM has multiple different causes which manifest in a similar pattern
" most patients are considered to be sporadic
" familial cases with dominant and recessive inheritance have been described
" in autosomal recessive for several families with different ethnic backgrounds have bee mapped to chromosome 9p1-q1
" possibly autoimmune T cell-mediated mechanism

### Clinical features
" onset age > 30 years, usually after 50 years
" duration of weakness >6 months
" muscle weakness affects proximal and distal muscles
" the patient must have at least one of the following:
    1. finger flexor weakness

2. wrist flexor weakness > wrist extensor weakness
3. quadriceps weakness >4 MRC scale

" dysphagia occurs in one third of patients
" muscles innervated by cranial nerve are mostly unaffected, but m.orbicularis oculi may be affected
" CK >12 times normal
" in sporadic IBM there is abnormal accumulation of beta amyloid protein and ubiquitin
" biopsy shows invasion of nonnecrotic muscle fibers by mononuclear cells, vacuolated muscle fibers, amyloid deposits or 15-18 nm tubolofilaments

**Strategy**

" demonstrate myopathy with spontaneous activity
" assess: severity: (mild, moderate, severe) and activity: (stationary, active)

**Expected abnormal findings**

*EMG*

" fibrillations
" small, brief MUPs

*Neurography*

" if distal muscles are severely involved MCS ampl are reduced

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.flexor carpi radialis/m.flexor digitorum profundus
" m.vastus lateralis/m.vastus medialis
" m.deltoideus/m.biceps
" m.interosseus dorsalis I/m.abductor digiti minimi
" m.tibialis anterior

*Neurography*

" MCS: n.medianus and n.peroneus unilaterally
" SCS: n.radialis and n.suralis unilaterally

**Note**

" MRI of the forearm muscles may be helpful in defining the pattern of affected flexor muscles, esspecially m.flexor digitorum profundus is affected

**References**


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**Modified**

" 26.6.1997 BF, 2.4.1997 BF, 3.4.97 ES

## 4.2 MUSCULAR DYSTROPHIES

### DUCHENNE MUSCULAR DYSTROPHY (DMD)

**Etiology**

" X-linked recessive inheritance
" mutation located at Xp21.2
" out of frame deletion of the dystrophin gene

" !/3 of the patients are new mutations
" 2/3 of the mothers to Duchenne boys are carriers of the gene

**Clinical features**

" incidence 1/3400 boys
" onset within the first 5 years
" delay in walking, abnormal gait, difficulty in climbing stairs
" proximal weakness, legs more than arms
" hypertrophy of calf muscles
" mild intellectual retardation
" loss of ambulation at 8-12 years
" life expectancy teens to 20 years
" CK very high

**Strategy**

" demonstrate myopathy with a proximal predominance

**Expected abnormal findings**

*EMG, myopathic usually with fibrillations*

" m.deltoideus/m.biceps
" m.vastus lateralis
" m.tibialis anterior

**Expected normal findings**

*Neurography*

" MCS
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

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**Modified**

" 30.3.1997, 3.4.97 ES

## BECKER MUSCULAR DYSTROPHY (BMD)

**Etiology**

" X-linked recessive inheritance
" mutation located at Xp21.2
" in frame deletion of the dystrophin gene

**Clinical features**

" onset variable, usually after 5 years may at 40 years or even later
" difficulty in running and climbing stairs
" proximal muscle weakness, initially only lower limb muscles
" quadriceps atrophy may be the only manifestation for a long time
" may present with cardiomyopathy
" calf hypertrophy
" slow progression
" ambulation after 16 years
" CK grossly elevated

**Strategy**

" demonstrate myopathy with a proximal predominance

**Expected abnormal findings**

*EMG, myopathic with fibrillations*

" m.deltoideus/m.biceps
" m.vastus lateralis
" m.tibialis anterior

**Expected normal findings**

*Neurography*

" MCS
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**


" Bushby KM, Goodship JA, Nicholson LV, Johnson MA, Haggerty ID, Gardner-Medwin D. Variability in clinical, genetic and protein abnormalities in manifesting carriers of Duchenne and Becker muscular dystrophy. Neuromusc Disord 1993,3:57-64
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**Modified**

" 30.3.1997 BF, 3.4.97 ES

## FACIO-SCAPULO-HUMERAL MUSCULAR DYSTROPHY (FSH)

**Etiology**

" autosomal dominant heredity
" linked to chromosome 4q35-4qter
" there is a deletion of an integral number of tandem repeats, there being a maximum of 8 of the original 12-96

**Clinical features**

" variable onset from childhood to adult age, 90 % will manifest before the age of 20
" an infantile variety has been described
" presenting symptom weakness of facial or shoulder muscles
" facial weakness is present in >50% of affected family members, particularly eye closure is affected
" scapular muscles are affected, particularly m.pectoralis major, however, m.deltoideus is spared for a long time
" asymmetry of the muscles is a rule, usually the right side is first affected
" some patients have weakness of the pelvic girdle
" course is variable: some may be mild with normal lifespan, some may loose ambulation in adult life
" cardiomyopathy is not a part of FSH
" CK normal or slightly elevated

**Strategy**

" demonstrate myopathic changes with facio-scapulo-humeralperoneal distribution

**Expected abnormal findings**

*EMG, myopathic findings in*

" m.trapezius
" m. infraspinatus
" m.tibialis anterior
" m.orbicularis oculi

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.biceps brachii/ m.triceps brachii
" m.infraspinatus/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" m.deltoideus may be normal in the early stages of FSH

**References**


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**Modified**

" 30.3.1997 BF, 3.4.97 ES

## EMERY-DREIFUSS MUSCULAR DYSTROPHY

**Etiology**

" X-linked recessive inheritance also autosomal dominant inheritance has been described with very similar clinical presentation

" in X-linked form the gene location is Xq27-28
" mutation in a gene called STA, which encodes a 254 amino acid protein, known as emerin. Emerin is expressed in most tissues, can be demonstrated from skin and leucocytes

**Clinical features**

" late childhood, adolescence, onset after 20 years is rare
" mild weakness
" focal wasting of m.biceps brachii and m.triceps brachii or m.gastrocnemii and the peroneal muscles
" rigidity of spine
" contractures of the elbow and the Achilles tendon develop before significant weakness
" equinus of the feet
" cardiac conduction defect, cardiomyopathy, cardiac arrhythmia life-threatening in early adult life
" slowly progressive
" CK moderately elevated but may be normal

**Strategy**

" demonstrate myopathic changes with humero-peroneal distribution, m.biceps brachii is particularly affected

**Expected abnormal findings**

*EMG, a mixture of small and large MUPs*

" m.biceps
" m.tibialis anterior
" paravertebral muscles in the lumbar and cervical region

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" contractures of joints often precede the weakness, especially in the elbow
" altered expression of emerin in patients and carriers can be tested with monoclonal antibodies from skin and leukocytes

**References**

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**Modified**

" 26.3.1997 BF, 3.4.97 ES, 21.4.1997 BF

## LIMB-GIRDLE MUSCULAR DYSTROPHY

**Etiology**

" heterogeneous
" most patients are autosomal recessive, two different forms have been found so far, one linked to chromosome 2p and another linked to chromosome 15q15
" calpain-3 deficiency and maps to 15q15
" autosomal dominant: gene located 5q22.1-31.3

**Clinical features**

" onset from early childhood to adult life
" difficulty with gait, running and climbing stairs
" onset with proximal muscle weakness in the pelvic or shoulder girdle muscles or both
" calf hypertrophy is common
" progression usually slow, but may sometimes be rapid
" CK elevated

**Strategy**

" demonstrate myopathic changes with limb-girdle distribution

**Expected abnormal findings**

*EMG*

" m.trapezius
" m.deltoideus
" m.vastus lateralis

*Neurography*

" MCS may show reduced amplitude in advanced cases

**Expected normal findings**

*Neurography*
" SCS
" MCS

**Procedure**

*EMG*
" m.deltoideus/m.biceps/m.trapezius
" m.interosseus/m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*
" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" Sometimes, m.extensor digitorum brevis is hypertrophic and may show high amplitude MUPs

**References**


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**Modified**

" 30.3.1997 BF, 3.4.97 ES

## CONGENITAL MUSCULAR DYSTROPHIES

" a group of myopathies often associated with central nervous system abnormalities, currently the group includes the following disorders.:
    1. Fukuyama congenital muscular dystrophy
    2. Walker-Warburg syndrome
    3. Muscle eye brain disease (MEB)
    4. Congenital muscular dystrophy with merosin deficiency
    5. Congenital muscular dystrophy with normal merosin
" Each of the disorders will be dealt with under its own heading.

## FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY

**Etiology**

" autosomal recessive inheritance, gene defect on chromosome 9q31-q33

**Clinical features**

" onset at birth, contractures, mean age of death around 8-10 years, most die by 18 years
" CNS involved, mental retardation, seizures
" no retinal abnormalities
" severe hypotonia, weakness and wasting of muscles
" seizures, mental retardation
" CK moderately high

**Strategy**

" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**

*EMG*
" myopathic findings in muscles, proximal muscles tend to be more involved

*Neurography*
" MCS amplitudes may be low

**Expected normal findings**

*Neurography*
" SCS
" MCS: CV and DLAT, F-responses

**Procedure**

*EMG*
" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*
" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Dubowitz V. 22nd ENMC sponsored workshop on congenital muscular dystrophy held in Baarn, The Netherlands, 14-16 May 1993. Neuromusc Disord 1994,4:75-82
" Fukuyama Y, Kawazura M, Haruna H. A peculiar form of congenital progressive muscular dystrophy. Report of fifteen cases. Paediatr Univ Tokyo 1960,4:5-8
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" Yoshioka M, Kuroki S. Clinical spectrum and genetic studies of Fukuyama congenital muscular dystrophy. Am J Med Genet 1994,53:245-250

**Modified**
" 30.3.1997, 3.4.97 ES

## WALKER-WARBURG SYNDROME

**Etiology**
" autosomal recessive inheritance
" deficiency of laminin β2 chain, linked to chromosome 3p21

**Clinical features**
" onset at birth
" mental retardation, type II lissencephaly, cerebellar and ocular malformations
" form of lissencephaly (type 2) with agyria
" an absent cortical layer
" absent or small corpus callosum and septum pellucidum
" small, dysplastic cerebellum and brain stem with absence of the posterior vermis
" CT scan will show a cobblestone appearance of the cortex and diffusely abnormal (cystic) white matter, as well as a marked area of translucency in the white matter around the ventricles.
" hydrocephalus has been variously attributed to aqueduct stenosis, Dandy-Walker malformation and herniation of the cerebellar tonsils
" retinal abnormalities
" CK variable
" death in infancy

**Strategy**
" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**
*EMG*
" myopathic findings in muscles, proximal muscles tend to be more involved
*Neurography*
" MCS amplitudes may be low

**Expected normal findings**
*Neurography*
" SCS
" MCS: CV and DLAT, F-responses

**Procedure**
*EMG*
" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis
*Neurography*
" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**
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" Denis D, Gambarelli D, Luciani A, et al. Walker-Warburg syndrome: a report of 3 cases. Ophthalmologica 1993,207:113-116
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" Kimura S, Sasaki Y, Kobayashi T, et al. Fukuyama-type congenital muscular dystrophy and the Walker-Warburg syndrome. Brain Dev 1993,15:182-191
" Warburg M. Hydrocephaly, congenital retinal nonattachment, and congenital falciform fold. Am J Ophthalmol 1978,85:88-94

**Modified**
" 30.3.1997 BF, 3.4.97 ES

## MUSCLE EYE BRAIN DISEASE (MEB)

**Etiology**
" recessive

**Clinical features**
" onset during the first six months of life
" slow motor development, severe mental retardation, hydrocephalus

" nystagmus and uncontrolled eye movements, anterior chamber defects with glaucoma, myopia, retinal dystrophy, cataracts and occasionally microspherophakia
" epilepsy common
" CK slightly elevated
" death by 6 to 16 years

**Strategy**

" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**

*EMG*

" myopathic findings in muscles, proximal muscles tend to be more involved

*Neurography*

" MCS amplitudes may be low

**Expected normal findings**

*Neurography*

" SCS
" MCS: CV and DLAT, F-responses

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Haltia M, Leivo J, Somer H, Pihko H, Pateau A, Kivelä T, Tarkkanen AS, Tomé F, Engvall E, Santavuori P. Muscle-Eye-Brain disease. A neuropathological study. Ann Neurol 1997;41:173-180
" Lenard HG. Congenital muscular dystrophies - problems of classification. Acta Paediatr Jpn 1991,33:256-260
" Pihko H, Lappi M, Raitta C, Sainio K, et al. Ocular findings in muscle-eye-brain (MEB) disease: a follow-up study. Brain Dev 1995,17:57-61
" Ranta S, Pihko H, Santavuori P, et al. Muscle-eye-brain disease and Fukuyama type congenital muscular dystrophy are not allelic. Neuromusc Disord 1995,5:221-225
" Santavuori P, Leisti J, Kruus S. Muscle, eye and brain disease. A new syndrome. Neuropediatrics 1977,8:550
" Santavuori P, Somer H, Sainio K, et al. Muscle-eye-brain disease (MEB). Brain Dev 1989,11:147-53
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" Warburg M. Muscle-eye-brain disease and Walker-Warburg syndrome: phenotype-genotype speculations (Commentary to Pihko's paper). Brain Dev 1995,17:62-63

**Modified**

" 30.3.1997 BF, 3.4.97 ES

## CONGENITAL MUSCULAR DYSTROPHY WITH MEROSIN DEFICIENCY

**Etiology**

" autosomal recessive inheritance, mutation of merosin (laminin alpa-2 chain) gene on chromosome 6q22-23

**Clinical features**

" neonatal hypotonia and muscle weakness
" never learn to walk
" MRI shows significant developmental CNS abnormalities
" intelligence often normal in spite of MRI abnormalities in the brain
" muscle histochemistry shows absence of merosin staining

**Strategy**

" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**

*EMG*

" myopathic findings in muscles, proximal muscles tend to be more involved

*Neurography*

" MCS amplitudes may be low

**Expected normal findings**

*Neurography*

" SCS
" MCS: CV and DLAT, F-responses

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Dubowitz V. 22nd ENMC sponsored workshop on congenital muscular dystrophy held in Baarn, The Netherlands, 14-16 May 1993. Neuromusc Disord 1994,4:75-82
" Fardeau M, Tome FMS, Helbling-Leclerc A, et al. Congenital muscular dystrophy with merosin deficiency: clinical, histopathological, immunocytochemical and genetic study. Rev Neurol 1996,152:11-19

" Hillaire D, Leclerc A, Faure S, Topaloglu H, et al. Localization of merosin-negative congenital muscular dystrophy to chromosome 6q2 by homozygosity mapping. Hum Molec Genet 1994,3:1657-1661
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**Modified**
" 30.3.1997 BF, 3.4.97

## CONGENITAL MUSCULAR DYSTROPHY WITH NORMAL MEROSIN

**Etiology**
" autosomal recessive inheritance

**Clinical features**
" neonatal hypotonia and muscle weakness
" non-progressive course
" mild disability, learn to walk in contrast to patients with merosin deficiency
" intelligence normal
" contractures
" CK variable, normal to moderately high
" normal merosin on muscle biopsy
" MRI normal CNS

**Strategy**
" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**
*EMG*
" myopathic findings in muscles, proximal muscles tend to be more involved
*Neurography*
" MCS amplitudes may be low

**Expected normal findings**
*Neurography*
" SCS
" MCS: CV and DLAT, F-responses

**Procedure**
*EMG*
" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis
*Neurography*
" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**
" Dubowitz V. 22nd ENMC sponsored workshop on congenital muscular dystrophy held in Baarn, The Netherlands, 14-16 May 1993. Neuromusc Disord 1994,4:75-82
" Kobayashi O, Hayashi Y, Arahata K, et al. Congenital muscular dystrophy: clinical and pathologic study of 50 patients with the classical (occidental) merosin-positive form. Neurology 1996,46:815-818
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" Topaloglu H, Kale G, Yalnizoglu D, et al. Analysis of "pure" congenital muscular dystrophies in thirty eight cases. How different is the classical type 1 from the occidental type cerebromuscular dystrophy?. Neuropediatrics 1994,25:94-100

**Modified**
" 30.3.1997 BF, 3.4.97

## OCULOPHARYNGEAL MUSCULAR DYSTROPHY

**Etiology**
" autosomal dominant inheritance with complete penetration
" located to chromosome 14q11.2

**Clinical features**
" onset during fourth to sixth decades
" ptosis, variable extraocular eye muscle weakness
" dysphagia
" mild proximal limb weakness
" CK normal or mildly elevated

**Strategy**
" demonstrate myopathic abnormalities in muscles innervated by cranial nerves
" differentiate from progressive external opthalmoplegia (mitochodrial myopathy)

**Expected abnormal findings**
*EMG*
" myopathic abnormalities
*Neurography*
" MCS amplitude may be reduced

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.orbicularis oculi, m.levator palpebrae
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Aarli JA. Oculopharyngeal muscular dystrophy. Acta Neurol Scand 1969,45:484-92
" Bouchard J-P, Gagne F, Tome F, Brunet D. The nuclear inclusions in oculopharyngeal muscular dystrophy in Quebec. Can J Neurol Sci 1989,16:446-450
" Brais B, Xie Y, Sanson M, et al. The oculopharyngeal muscular dystrophy locus maps to the region of the cardiac alpha and beta myosin heavy chain genes on chromosome 14q. Can J Neurol Sci 1995,22 suppl 1:S17 F1
" Wong KT, Dick D, Anderson JR. Mitochondrial abnormalities in oculopharyngeal muscular dystrophy. Neuromusc Disord 1996,6:163-166

**Modified**

" 30.3.1997 BF, 3.4.97 ES

### PRIMARY ADHALINOPATHY (α-SARCOGLYCAGONAPATHY)

**Etiology**

" autosomal recessive inheritance with complete penetration
" mapped to chromosome 17q12-q21 and in some patients to 13q12
" different subtypes of primary adhalinopathy have been described
" also secondary adhalinopathies exist

**Clinical features**

" clinically similar to Duchenne dystrophy
" onset during average onset around 8 years, ranging from 3 to 15 years
" weakness of proximal muscles most in the pelvic girdle muscles
" distal muscle involvement in the early stages is minimal, m.tibialis anterior predominates
" trunk extensor more affected than abdominal muscles
" neck muscles spared
" calf hypertrophy common
" CK elevated

**Strategy**

" demonstrate myopathic abnormalities mainly in proximal limb and trunk muscles

**Expected abnormal findings**

*EMG*

" myopathic abnormalities

*Neurography*

" MCS amplitude may be reduced

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.orbicularis oculi, m.levator palpebrae
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Eymard B, Romero NB, Leturc F, Piccolo F, Carrié A ;Jeanpierre M, Collin H, Debyrgrave N, Azibi K et al, Primary adhalinopathy (α-sarcoglycanopathy): clinical, pathologic and genetic correlations in 20 patients with autosomal recessive muscular dystrophy. Neurology 1997; 48:1227-1234

**Modified**

" 26.6.1997 BF

## 4.3 DISTAL MYOPATHIES

### LATE ONSET DISTAL MYOPATHY TYPE 1 (WELANDER TYPE)

**Etiology**

" autosomal dominant heredity

**Clinical features**

" onset around 45-50 years (range 20 to 80 years)
" distal hand muscle weakness, especially extension of the thumb and index finger
" distal leg muscles are affected later (in contrast to tibial muscle dystrophy which starts in the legs)
" slow progression, normal life expectancy
" distal tendon reflexes are often absent
" CK either normal or mildly elevated
" most patients described have Swedish ancestry
" in Sweden this myopathy is common around 100 km north from Stockholm in a village called Hedesunda. Local people call this distal myopathy "Hedesunda disease"

**Strategy**

" demonstrate myopathic changes with distal predominance in arms and legs

**Expected abnormal findings**

*EMG*

" m.interosseus dorsalis
" m.extensor digitorum communis
" m.tibialis anterior

**Expected normal findings**

*EMG*

" m.biceps brachii
" m.vastus lateralis/m.vastus medialis

*Neurography*

" SCS
" MCS may show reduced amplitudes

**Procedure**

*EMG*

" m.biceps
" m.interosseus
" m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" EMG often shows fibrillation potentials and complex repetitive discharges. In severely affected muscles reduced interference pattern may be seen.

**References**

" Ahlberg G, Jakobsson F, Fransson A, Moritz A, et al. Distribution of muscle degeneration in Welander distal myopathy - a magnetic resonance imaging and muscle biopsy study. Neuromusc Disord 1994;4:55-62
" Borg K, Tome FMS, Edstrom L. Intranuclear and cytoplasmic filamentous inclusions in distal myopathy (Welander). Acta Neurol Scand 1991;82:102-106
" Edstrom L. Histochemical and histopathological changes in skeletal muscle in late-onset hereditary distal myopathy (Welander). J Neurol Sci 1975;47:147-57
" Welander L. Myopathia distalis tarda hereditaria. Acta Medica Scand 1951;141:1-124

**Modified**

" 1.4.1997 BF, 3.4.97 ES

## LATE ONSET DISTAL MYOPATHY TYPE 2 (MARKESBURY)

**Etiology**

" autosomal dominant heredity

**Clinical features**

" starts with weakness of distal leg muscles and spreads later to hand muscles
" late in the course also proximal limb muscles are involved
" cardiomyopathy with congestive heart failure and tachyarrytmias may occur
" CK normal or slightly elevated

**Strategy**

" demonstrate myopathic changes with distal predominance
" legs affected more than arms

**Expected abnormal findings**

*EMG*

" m.tibialis anterior
" m.interosseus dorsalis
" m.extensor digitorum communis
" m.deltoideus/m.biceps (later in the disease)
" m.vastus lateralis (later in the disease)

**Expected normal findings**

*EMG (in the early stages of the disease)*

" m.biceps brachii
" m.vastus lateralis/m.vastus medialis

*Neurography*

" SCS
" MCS may show reduced amplitudes

**Procedure**

*EMG*

" m.biceps
" m.interosseus
" m.extensor digitorum communis

" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Markesbery WR, Griggo RC, Herr B. Distal myopathy: electron microscopic and histochemical studies. Neurology 1977;27:727-35

**Modified**

" 1.4.1997 BF, 3.4.97 ES

## EARLY ADULT ONSET DISTAL MYOPATHY TYPE 1

**Etiology**

" autosomal recessive or sporadic
" some cases may be identical with hereditary inclusion body myopathy

**Clinical features**

" onset with weakness of the muscles in the anterior compartment of the leg
" slow progression
" hand muscles affected
" proximal limb muscles may be affected later in the disease

**Strategy**

" demonstrate myopathic changes with distal predominance
" legs affected more than arms

**Expected abnormal findings**

*EMG*

" m.tibialis anterior
" m.interosseus dorsalis
" m.extensor digitorum communis
" m.deltoideus/m.biceps (later in the disease)
" m.vastus lateralis (later in the disease)

**Expected normal findings**

*EMG (in the early stages of the disease)*

" m.biceps brachii
" m.vastus lateralis/m.vastus medialis

*Neurography*

" SCS
" MCS may show reduced amplitudes

**Procedure**

*EMG*

" m.biceps
" m.interosseus
" m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Nonaka I, Sunohara N, Satoyoshi E, et al. Autosomal recessive distal muscular dystrophy. Ann Neurol 1985;17:51-59

**Modified**

" 1.4.1997, 3.4.97 ES

## EARLY ADULT ONSET DISTAL MYOPATHY TYPE 2 (MIYOSHI)

**Etiology**

" autosomal recessive or sporadic
" linked to chromosome 2p12-14

**Clinical features**

" onset second or third decade
" onset of weakness and atrophy of the calf muscles
" usually sparing of intrinsic foot muscles
" severe progressive disorder, results in loss of ambulation
" CK moderately to severely elevated (10 to 150 times normal)

**Strategy**

" demonstrate myopathic changes with distal predominance
" legs affected more than arms

**Expected abnormal findings**

*EMG*

" m.gastrocnemius >m.tibialis anterior
" m.vastus lateralis (later in the disease)

**Expected normal findings**

*Neurography*

" SCS
" MCS may show reduced amplitudes

**Procedure**

*EMG*
" m.biceps
" m.interosseus
" m.extensor digitorum communis
" m.vastus lateralis
" m.tibialis anterior

*Neurography*
" SCS: n.suralis, n.radialis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Barohn RJ, Miller RG, Griggs RC. Autosomal recessive distal dystrophy. Neurology 1991;41:1365-1370
" Bejaoui K, Hirabayashi K, Hentati F, et al. Linkage of Miyoshi myopathy (distal autosomal recessive muscular dystrophy) locus to chromosome 2p12-14. Neurology 1995;45:768-772
" Miyoshi K, Kawai H, Iwasa M, et al. Autosomal recessive distal muscular dystrophy as a new type of progressive muscular dystrophy: seventeen cases in eight families including an autopsied case. Brain 1986;109:31-54
" Yamanouchi Y, Ozawa E, Nonaka I. Autosomal recessive distal muscular dystrophy: normal expression of dystrophin, utrophin and dystrophin-associated proteins in muscle fibers. J Neurol Sci 1994;126:70-76


**Modified**
" 1.4.1997, 3.4.97 ES

### TIBIAL MUSCULAR DYSTROPHY

**Etiology**
" autosomal dominant heredity

**Clinical features**
" peroneal weakness, starting at the age 35 or later
" hand muscles appear clinically normal
" the muscle abnormalities are most prominent in the muscles peroneal muscles of the leg, however m.extensor digitorum brevis in the foot is spared
" hamstring muscles and calf muscles may be mildly affected
" CK is normal or mildly elevated
" slow progression

**Strategy**
" demonstrate myopathic changes with distal predominance in the lower extremeties

**Expected abnormal findings**
*EMG*
" tibialis anterior, myopathic

**Expected normal findings**
*EMG*
" m.biceps brachii/m.deltoideus
" m.vastus lateralis/m.vastus medialis
" m.extensor digitorum brevis shows normal age related abnormalities

*Neurography*
" all nerves, also n.peroneus (m.extensor digitorum brevis is spared).

**Procedure**
*EMG*
" m.interosseus dorsalis I
" m.tibialis anterior
" m.extensor digitorum brevis
" m.deltoideus/m.biceps brachii
" m.vastus lateralis

*Neurography*
" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**
" may show EMG complex repetitive discharges. in severely affected muscles reduced interference pattern may be seen.

**References**

" Udd B, Partanen J, Halonen P, Falck B, et al. Tibial muscular dystrophy. Late adult-onset distal myopathy in 66 Finnish patients. Arch Neurol 1993,50:604-608
" Nokelainen P, Udd B, Somer H, Peltonen L. Linkage analysis in tibial muscular dystrophy. Hum Hered 1996,46:98-107
" Partanen J, Laulumaa V, Paljarvi L, et al. Late onset foot-drop muscular dystrophy with rimmed vacuoles. J Neurol Sci 1994,125:158-167


**Modified**
" 30.3.1997, 3.4.97 ES

## 4.4 MYOTONIAS

### MYOTONIC DYSTROPHY

**Etiology**
" autosomal dominant inheritance
" gene location 19q13.2-q13.3
" there is an expanded CGT trinucleotide repeat. In normal subjects the length is up to 30 repeats in patients with myotonic dystrophy up to 2000 repeats can be found. Severity is related with the number of repeats

" gene product myotonin

**Clinical features**

" myotonic dystrophy is the most common adult myopathy, prevalence is around 5/100 000
" presentation and onset age varies considerably
" onset usually in the early teens with distal muscle weakness in arms and legs
" sometimes myotonic dystrophy presents in a congenital form with hypotonia, bilateral facial weakness, mental retardation, neonatal respiratory distress and talipes; the mother is almost invariably a carrier of the myotonic dystrophy gene in congenital myotonic dystrophy
" facial muscles and sternocleiodomastoid muscles are also weak, mild ptosis
" myotonia is usually present and can be demonstrated in the hand muscles
" patients very rarely complain about myotonia (unlike patients with myotonia congenita)
" myotonia is reduced after exercise "warm-up" effect like myotonia congenita.
" myotonic dystrophy affects many other organ systems:
    6. cataracts, retinal dysfunction
    7. CNS, mild mental dysfunction
    8. premature balding
    9. mild polyneuropathy is sometimes seen
    10. endocrine abnormalities
    11. smooth muscle dysfunction

**Strategy**

" demonstrate myotonic discharges
" demonstrate myopathic abnormalities with a distal distribution

**Expected abnormal findings**

*EMG*

" in distal muscles usually abundant myotonic discharges in adults
" trains of positive waves, sometimes difficult to differentiate from myotonic discharges
" short duration, polyphasic MUPs in distal muscles
" typically dense interference pattern, maybe reduced in advanced stages

*Neurography*

" *MCV* mostly normal CV, in 10 % mild to moderate reduced CV, sometimes low amplitude
" *SCS n*ormal

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.extensor digitorum communis
" m.masseter/m.orbicularis oris/m. sternocleidomastoideus
" m.deltoideus/m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" some adults (very rarely) with myotonic dystrophy do not have myotonic discharges
" generally children with myotonic dystrophy do not show myotonic discharges
" children with congenital myotonic dystrophy do not show myotonic discharges or any significant abnormalities on EMG
" if congenital myotonic dystrophy is suspected, study also the mother, in almost all children with the congenital form the mother is affected
" around 10 % of patients with myotonic dystrophy have a mild polyneuropathy

**References**


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**Modified**

" 28.3.1997 BF, 3.4.97 ES

## PROXIMAL MYOTONIC MYOPATHY (PROMM)

**Etiology**

" autosomal dominant inheritance

**Clinical features**

" onset of symptoms at the age 20 to 60 years
" myotonia varies over time, may be absent on many days, myotonia has a warm-up effect
" electrical myotonia decreases with cold and increases after heating
" mild proximal weakness especially in the legs
" clinical weakness tends to be proximal but electrophysiological abnormalities are more pronounced distally
" muscle pain is common often disabling
" often polychromatic posterior cataracts

" diabetes is common
" clinical course is mild

**Strategy**

" demonstrate myotonia, myopathic MUP abnormalities with a proximal preponderance
" differentiate from myotonic dystrophy

**Expected abnormal findings**

*EMG*

" myotonic discharges, especially in distal muscles
" MUP analysis normal or mild myopathic abnormalities, especially in distal muscles

**Expected normal findings**

*Neurography*

" MCS
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

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**Revisions**

" 16.1.1997 BF, 28.3.1997 BF, 3.4.97 ES

## MYOTONIA CONGENITA (THOMSEN´S FORM)

**Etiology**

" autosomal dominant inheritance
" gene location 7q35
" gene product: muscle $Cl^-$ channel, which are formed of tetrameres; the mutation interferes with tetramere formation
" the chloride conductance in the muscle fibre membrane is reduced resulting in accumulation of $K^+$ ions in the t-tubules
" the membrane depolarization is prolonged giving rise to spontaneous repeated action potentials

**Clinical features**

" onset in infancy
" myotonia is detected in childhood and persists throughout life
" presentation varies considerably even within families, penetrance aroun 90%
" myotonia is usually mild
" myotonia decreases after repeated movements, “warm-up”
" muscle strength normal
" often muscles appear hypertrophic
" CK normal or occasionally borderline
" myotonia may aggravated by $\beta_2$ agonist drugs

**Strategy**

" demonstrate myotonic discharges without other myopathic features in muscles

**Expected abnormal findings**

*EMG*

" myotonic discharges, particularly distally (sometimes after provocation such as percussion, needle movements, cooling)
" warming up effect - less myotonia after a period of maximal contraction

*Repetitive nerve stimulation*

" decrementing response, especially at high stimulation frequencies (30 Hz) without intratetanic facilitation
" following exercise the amplitude is decreased, reverse to facilitation, cooling does not affect this in contrast to paramyotonia congenita

**Expected normal findings**

*Neurography*

" normal

*EMG*

" MUPs are usually normal

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.extensor digitorum communis
" m.masseter/m.orbicularis oris

" m.deltoideus/m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**


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**Modified**

" 27.3.1997, 3.4.97 ES

## MYOTONIA CONGENITA (BECKER´S FORM)

**Etiology**

" autosomal recessive inheritance
" gene location chromosome 7q35
" gene product:: muscle chloride channel

**Clinical features**

" myotonia manifests at 7-14 years of age, sometimes later and persists throughout life
" myotonia in Becker's form is more severe than Thomsen's form
" muscle strength is normal, on exercise strength decreases
" leg and gluteal muscles are usually hypertrophic
" weakness is more prominent in the arms and stiffness in the legs
" lordosis is common
" neck, shoulder and arm muscles appear atrophic
" myotonia decreases after repeated movements, "warm-up"
" male carriers are said to have myotonia, but not females
" CK normal or mildly elevated (2-3 times normal)

**Strategy**

" demonstrate myotonic discharges, in many patients EMG shows distally mild myopathic features

**Expected abnormal findings**

*EMG*

" myotonic discharges, particularly distally
" MUP analysis often shows mild myopathic abnormalities
" warming up effect - less myotonia after a period of maximal contraction

*Repetitive nerve stimulation*

" decrementing response, especially at high stimulation frequencies (30 Hz)
" following exercise the amplitude is decreased, reverse to facilitation
" cooling does not affect this in contrast to paramyotonia congenita

**Expected normal findings**

*Neurography*

" normal

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.extensor digitorum communis
" m.masseter/m.orbicularis oris
" m.deltoideus/m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" male carriers show myotonic discharges on EMG, but not female carriers

**References**


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**Modified**

" 28.3.1997, 3.4.97 ES

## MYOTONIA FLUCTUANS

**Etiology**
- autosomal dominant inheritance
- gene location chromosome 17q13,1
- mutation of muscle alpha-subunit of the sodium channel
- genetic defect allelic to hyperkalemic periodic paralysis and paramyotonia congenita

**Clinical features**
- myotonia is precipitated by exercise, develops during rest within 30 minutes and lasts for about one hour
- at rest there is no myotonia
- normal muscle strength
- cold does not precipitate myotonia like in paramyotonia congenita
- potassium loading aggravates myotonia
- depolarizing muscle relaxants (e.g. suxamethonium) exacerbates myotonia

**Strategy**
- demonstrate myotonic discharges at rest with lack of structural myopathic abnormalities
- differentiate from paramyotonia congenita, myotonia congenita and hyperkalemic periodic paralysis

**Expected abnormal findings**

*EMG*
- myotonic discharges
- there may fibrillation like spontaneous activity at room temperature
- following exercise the amount myotonic discharges increase

**Expected normal findings**

*EMG*
- MUP analysis

*Neurography*
- MCS
- SCS

**Procedure**

*EMG*
- m.deltoideus/m.biceps brachii
- m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
- m.vastus lateralis/m.vastus medialis/m.tensor facia latae
- m.tibialis anterior

*Neurography*
- SCS: n.suralis and n.radialis on one side
- MCS: n.peroneus and n.medianus on one side

**References**

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**Modified**
- 28.3.1997, 3.4.97 ES

## PARAMYOTONIA CONGENITA

**Etiology**
- autosomal dominant inheritance
- gene location chromosome 17q13,1
- mutation of muscle alpha-subunit of the sodium channel
- genetic defect allelic to hyperkalemic periodic paralysis and myotonia fluctuans

**Clinical features**
- symptoms present from birth and persist throughout life
- myotonia is paradoxical, it increases during exercise
- severe worsening of exercise induced myotonia in cold
- predilection of face, neck and distal upper extremity muscles
- weakness after prolonged exercise and cold in most cases
- CK elevated up to 5-10 times normal

**Strategy**
- demonstrate myotonic discharges, in many patients EMG shows distally mild myopathic features

**Expected abnormal findings**

*EMG*
- myotonic discharges, much less prominent than in other myotonias
- cooling will initially induce repetitive spontaneous motor unit discharges, some authors describe fibrillation like activity in cooled muscles
- with increased cooling myotonia disappears with complete muscle depolarization and paralysis

*Neurography*
- MCS amplitude is reduced in cooled muscles

**Expected normal findings**

*Neurography*
- normal

**Procedure**

*EMG*
- m.interosseus dorsalis I
- m.tibialis anterior

" m.extensor digitorum communis
" m.masseter/m.orbicularis oris
" m.deltoideus/m.vastus lateralis

*Muscle cooling*

" cooling of hand muscles

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**


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**Modified**

" 28.3.1997 BF, 3.4.97 ES

## CHONDRODYSTROPHIC MYOTONIA (SCHWARTZ-JAMPEL SYNDROME)

**Etiology**

" autosomal recessive inheritance
" localized to 1p34-p36

**Clinical features**

" small stature, multiple skeletal deformities and myopia
" kyphoscoliosis, lumbar lordosis, pectus carinatum, bowing of the long bones, pes planus, a valgus deformity of the ankles and wide metaphyses.
" radiographs show platyspondyly, coronal clefts of the vertebral bodies, and an epiphyseal dysplasia, especially around the hip
" muscle stiffness
" symptoms appear within the first 3 years
" thigh muscles appear hypertrophic, shoulder girdle muscles atrophic
" non-progressive

**Strategy**

" demonstrate continuos spontaneous activity with little modulation of amplitude and frequency (complex repetitive discharges) some authors also describe myotonic discharges. Sometimes this is best seen in m. orbicularis oris.

**Expected abnormal findings**

" myotonic discharges or complex repetitive present from age 7 months onwards

**Expected normal findings**

*Neurography*

" normal SCS
" normal MCS

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.extensor digitorum communis
" m.masseter/m.orbicularis oris
" one proximal muscle: m.deltoideus or m.quadriceps femoris (may show only little abnormalities)

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**Note**

" the children have arthrogryposis, kyfoscoliosis, pectus carinatum and short stature

**References**


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**Modified**

" 12.5.1977 BF, 28.3.1997 BF, 3.4.97 ES

# 4.5 CONGENITAL MYOPATHIES

## CENTRAL CORE DISEASE

**Etiology**
" autosomal dominant inheritance
" gene location chromosome 19q12-13.1 (allelic to malignant hypertermia)
" mutation of ryanodine receptor

**Clinical features**
" onset in infancy, rarely later
" hypotonia, proximal muscles > distal, legs>arms
" extraocular muscles spared, facial muscles little involved
" slowly progressive
" kyphoscoliosis, congenital hip dislocation common, pes cavus
" characteristic histological findings
" CK: high
" slowly progressive

**Strategy**
" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**
*EMG*
" myopathic findings in muscles, proximal muscles tend to be more involved

**Expected normal findings**
*Neurography*
" SCS
" MCS

**Procedure**
*EMG*
" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis
*Neurography*
" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**
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**Modified**
" 1.4.1997 BF, 3.4.97 ES

## NEMALINE MYOPATHY

**Etiology**
" genetically heterogeneous
" autosomal dominant inheritance common: gene defect localized to chromosome 1q21-23 (mild form)
" autosomal recessive inheritance: chromosome 2q21.2-q22 (severe form)

**Clinical features**
" onset in infancy, hypotonia, proximal muscles > distal
" the weakest muscles were the facial muscles, flexors of the neck and trunk, dorsiflexors of the feet and the extensors of the toes
" extraocular muscles spared
" proximal and distal muscles affected
" no artrogroposis
" pectus excavatum, kyphoscoliosis, talipes equinovarus
" no cardiac involvement
" nemaline bodies on Gomori trichrome staining
" CK usually normal or slightly elevated

**Strategy**
" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**
*EMG*
" myopathic findings in muscles, proximal muscles tend to be more involved, EMG may be normal

**Expected normal findings**
*Neurography*
" SCS
" MCS

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**


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"

**Modified**

" 1.4.1997, BF, 3.4.97

## CENTRONUCLEAR MYOPATHY (MYOTUBULAR MYOPATHY) NEONATAL FORM

**Etiology**

" X-linked recessive inheritance
" gene location Xq28

**Clinical features**

" neonatal onset
" polyhydramnion
" narrow face and long fingers
" severe hypotonia and weakness
" respiratory insufficiency
" swallowing difficulties
" opthalmoplegia and ptosis
" fatal outcome, mean life expectancy 5 months
" normal or mild elevation of CK

**Strategy**

" demonstrate myopathic abnormalities

**Expected abnormal findings**

*EMG*

" fibrillations and positive sharp waves, complex repetitive discharges may be seen
" MUP analysis shows myopathic abnormalities

*Neurography*

" MCS may show reduced amplitude

**Expected normal findings**

*Neurography*

" SCS
" MCS normal CV, DLAT, decay and F Waves

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**


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**Modified**
" 1.4.1997, 3.4.97 ES

## CENTRONUCLEAR MYOPATHY (MYOTUBULAR MYOPATHY) LATE INFANTILE-LATE CHILDHOOD FORM

**Etiology**
" autosomal recessive inheritance

**Clinical features**
" onset late infancy to childhood
" hypotonia
" ptosis, ophtalmoplegia
" skeletal abnormalities
" areflexia
" slowly progressive
" normal or mild elevation of CK

**Strategy**
" demonstrate myopathic abnormalities

**Expected abnormal findings**
*EMG*
" fibrillations and positive sharp waves, complex repetitive discharges may be seen
" MUP analysis shows myopathic abnormalities
*Neurography*
" MCS may show reduced amplitude

**Expected normal findings**
*Neurography*
" SCS
" MCS normal CV, DLAT, decay and F Waves

**Procedure**
*EMG*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**
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" Heckmatt JZ, Sewry CA, Hodes D, Dubowitz V. Congenital centronuclear (myotubular) myopathy: a clinical, pathological and genetic study in eight children. Brain 1985;108:941-964
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" Wallgren-Pettersson C, Thomas NST. Report on the 20th ENMC sponsored international workshop: myotubular/centronuclear myopathy. Neuromusc Disord 1994;4:71-74
" Wallgren-Pettersson C, Clarke A, Samson F, Fardeau M, et al. The myotubular myopathies: differential diagnosis of the X linked recessive, autosomal dominant, and autosomal recessive forms and present state of DNA studies. J Med Genet 1995;32:673-679

**Modified**
" 1.4.1997 BF, 3.4.97 ES

## CENTRONUCLEAR MYOPATHY (MYOTUBULAR MYOPATHY) LATE CHILDHOOD-ADULT TYPE

**Etiology**
" autosomal dominant inheritance

**Clinical features**
" onset late childhood to adult age
" mild limb-girdle weakness
" facial and external ocular muscles may be affected
" normal or mild elevation of CK
" slowly progressive

**Strategy**
" demonstrate myopathic abnormalities

**Expected abnormal findings**
*EMG*
" fibrillations and positive sharp waves, complex repetitive discharges may be seen
" MUP analysis shows myopathic abnormalities
*Neurography*
" MCS may show reduced amplitude

**Expected normal findings**

*Neurography*

" SCS
" MCS normal CV, DLAT, decay and F-waves

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**


" De Angelis MS, Palmucci L, Leone M, et al. Centronuclear myopathy: clinical, morphological and genetic characters. A review of 288 cases. J Neurol Sci 1991;103:2-9
" Goebel HH, Meinck HM, Reinecke M, et al. Centronuclear myopathy with special consideration of the adult form. Eur Neurol 1984;23:425-34
" McLeod JG, Baker De C, Lethlean AK, et al. Centronuclear myopathy with autosomal dominant inheritance. J Neurol Sci 1972;15:375-7
" Wallgren-Pettersson C, Thomas NST. Report on the 20th ENMC sponsored international workshop: myotubular/centronuclear myopathy. Neuromusc Disord 1994;4:71-74
" Wallgren-Pettersson C, Clarke A, Samson F, Fardeau M, et al. The myotubular myopathies: differential diagnosis of the X linked recessive, autosomal dominant, and autosomal recessive forms and present state of DNA studies. J Med Genet 1995;32:673-679


**Modified**

" 1.4.1997 BF, 3.4.97 ES

## MULTICORE DISEASE

**Etiology**

" probably autosomal dominant inheritance

**Clinical features**

" delayed motor milestones
" proximal muscle weakness
" rarely ptosis and opthalmoplegia
" skeletal abnormalities (slender body, high arched plate, clubfeet, scoliosis)
" predisposition to malignant hypertermia
" non-progressive
" CK normal

**Strategy**

" demonstrate myopathic abnormalities
" EMG findings not specific

**Expected abnormal findings**

*EMG*

" variable degrees of myopathic abnormalities

*Neurography*

" MCS may show reduced amplitude

**Expected normal findings**

*Neurography*

" SCV
" MCV: nerve conduction velocity, decay and F-waves

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**


" Shuaib A, Martin JME, Mitchell LB, Brownell AKW. Multicore myopathy: not always a benign entity. Can J Neurol Sci 1988;15:10-14


**Modified**

" 1.4.1997, 3.4.97 ES

## MINI CORE DISEASE

**Etiology**

" sporadic or autosomal recessive

**Clinical features**

" onset in infancy rarely later
" onset in infancy, hypotonia, proximal muscles > distal
" ptosis, extraocular muscles weak
" cardiac involvement
" non progressive

" distinctive electron microscopy findings

**Strategy**

" demonstrate myopathic abnormalities, sometimes EMG may be normal
" differentiate from SMA1

**Expected abnormal findings**

*EMG*

" myopathic findings in muscles, proximal muscles tend to be more involved

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Aaberg TM, Yip WCL. A fatal congenital myopathy with severe type I fibre atrophy, central nuclei and multicores. J Neurol Sci 1981;50:277-90
" Lake BD, Wilson J. Myopathy with minicore in siblings. Neuropath Appl Neurobiol 1977;3:159-67
" Shy GM, Magee KR. A new congenital non-progressive myopathy. Brain 1956;79:610-621

**Modified**

" 1.4..1997, 3.4.97 ES

## FINGERPRINT BODY MYOPATHY

**Etiology**

" pathogenesis is unknown
" rare
" not certain that this is a separate entity, fingerprint like structures can be seen in other myopathies

**Clinical features**

" hypotonia in infancy
" truncal and extremity weakness
" cranial muscles spared
" non-progressive or slowly progressive
" mental retardation
" electron microscopy shows typical fingerprint patterns

**Strategy**

" demonstrate myopathic abnormalities
" differentiate from SMA1

**Expected abnormal findings**

*EMG*

" myopathic findings

**Expected normal findings**

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.interosseus dorsalis I
" m.tibialis anterior
" m.deltoideus
" m.vastus lateralis

*Neurography*

" SCS: n.radialis and n.suralis unilaterally
" MCS: n.medianus and n.peroneus unilaterally

**References**

" Engel AG, Gordon AS, Rewcastle NB, et al. Chronic benign congenital myopathy: fingerprint body type. Can J Neurol Sci 1974;1:106-113
" Fardeau M, Tome FMS, Derambure S. Familial fingerprint body myopathy. Arch Neurol 1976;33:724-5

**Modified**

" 1.4.1997, 3.4.97 ES

## HYALINE BODY MYOPATHY

**Etiology**

" sporadic occurrence
" etiology not known

**Clinical features**

" hypotonia
" delayed milestones
" weakness of proximal muscles

" slow progression or stationary
" CK normal

**Strategy**

" demonstrate myopathic abnormalities in more severe proximal than in distal muscles

**Expected abnormal findings**

*EMG*

" myopathic abnormalities

*Neurography*

" MCS may show reduced amplitudes

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Barohn RJ, Brumback RA, Mendell JR. Hyaline body myopathy. Neuromusc Disord 1994;4:257-262
" Ceuterick C, Martin JJ, Martens C. Hyaline bodies in skeletal muscle of a patient with a mild chronic non-progressive congenital myopathy. Clin Neuropathol 1993;12:79-83

**Modified**

" 1.4.1997 BF, 3.4.97 ES

## 4.6 MITOCHONDRIAL MYOPATHIES

### MYOCLONIC EPILEPSY AND RAGGED RED FIBERS (MERFF)

**Etiology**

" maternal inheritance
" point mutation of mitochondrial DNA, an adenine to guanine substitution at nucleotide 8344 in the transfer-RNA gene for lysine
" the mitochondrial DNA population is heteroplasmic, there is mutant and wild type DNA is varying proportions

**Clinical features**

" early development is usually normal
" onset of symptoms varies from childhood to adult age (5 to 50 years)
" myopathy with proximal weakness
" myoclonus
" generalized seizures
" ataxia
" hearing loss and deafness
" intellectual deterioration
" CK normal or mildly elevated
" muscle biopsy shows ragged red fibers on Gomori trichrome staining
" electron microscopy shows abnormal mitochondria
" some patients have neuropathy

**Strategy**

" demonstrate myopathic abnormalities in proximal muscles
" EEG shows generalized slowing and generalized spike and wave discharges

**Expected abnormal findings**

*EMG*

" MUP analysis shows myopathic abnormalities

*Neurography*

" may show slight abnormalities

**Expected normal findings**

*Neurography*

" may be normal

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

*EEG*

**References**

" Acharya JN, Satishchandra P, Shankar SK. Familial progressive myoclonus epilepsy: clinical and electrophysiologic observations. Epilepsia 1995,36:429-34

" Berkovic SF, Carpenter S, Evans A, et al. Myoclonus epilepsy and ragged-red fibres (MERRF). I. A clinical, pathological, biochemical, magnetic resonance spectrographic and positron emission tomographic study. Brain 1989,112:1231-1260
" Franceschetti S, Antozzi C, Binelli S, Carrara F, Nardocci N, Zeviani M, Avanzini G. Progressive myoclonus epilepsies: an electroclinical, biochemical and genetic study. Acta Neurol Scand 1993,87:219-23
" Fukuhara N. Clinicopathological features of MERRF. Muscle Nerve 1995,Suppl 3:S90-4
" Serra G, Piccinnu R, Tondi M, et al. Clinical and EEG findings in eleven patients affected by mitochondrial encephalomyopathy with MERRF - MELAS overlap. Brain Dev 1996,18:185-191
" Suomalainen A, Ciafaloni E, Koga Y, et al. Use of single strand conformation polymorphism analysis to detect point mutations in human mitochondrial DNA. J Neurol Sci 1992,111:222-226

**Modified**

" 1.4.1997, 3.4.97 ES

## MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS AND STROKELIKE EPISODES (MELAS)

**Etiology**

" may be familial, but far less often than MERRF
" point mutation of mitochodrial DNA, an adenine to guanine substitution occurs in the tRNA for leucine at nucleotide 3243.
" the mutant DNA coexists with normal DNA

**Clinical features**

" early development normal
" begins usually between 3 and 11 years, before 20 years
" stroke like episodes
" seizures
" short stature
" episodic vomiting
" hearing loss
" intellectual deterioration
" myopathy
" CK may be elevated
" lactate, especially CSF lactate elevated

**Strategy**

" evaluate presence of myopathic abnormalities

**Expected abnormal findings**

*EMG*

" may show mild myopathic abnormalities

*EEG*

" generalized slowing
" focal abnormalities may be seen
" focal and generalized epileptiform discharges

**Expected normal findings**

*EMG*

" may be normal

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

*EEG*

**References**

" Anonymous. Mitochondrial encephalomyopathies: gene mutation. Neuromusc Disord 1994;4:397-398
" Ciafaloni E, Ricci E, Shanske S, et al. MELAS: Clinical features, biochemistry, and molecular genetics. Ann Neurol 1992;42:391-398
" Damian MS, Seibel P, Reichmann H, et al. Clinical spectrum of the MELAS mutation in a large pedigree. Acta Neurol Scand 1995;92:409-415
" Goto Y. Clinical features of MELAS and mitochondrial DNA mutations. Muscle Nerve 1995;Suppl 3:S107-12
" Harding AE. Mitochondrial genes and neurological disease. Clin Exp Neurol 1993;30:1-16
" Suomalainen A, Majander A, Pihko H, Peltonen L, Syvanen A-C. Quantification of tRNA(3243 Leu) point mutation of mitochondrial DNA in MELAS patients and its effects on mitochondrial transcription. Hum Mol Genet 1993;2:525-534

**Modified**

" 1,4,1997, 3.4.97 ES

## SUCCINATE DEHYDROGENASE DEFICIENCY (COMPLEX II DEFICIENCY)

**Etiology**

" Succinate dehydrogenase is coded by nuclear DNA, autosomal recessive inheritance has been described

**Clinical Features**

" presents in childhood with exercise intolerance
" rhabdomyolysis and myoglobinuria may be present
" between attacks normal neurologic examination

" in relation to attacks elevated CK and abnormal EMG
" cardiac muscle spared

**Strategy**

" usually normal EMG between attacks
" demonstrate abnormal EMG in relation to attacks

**Expected abnormal findings in relation to attacks**

*EMG*

" fibrillations and positive sharp waves
" MUP analysis myopathic

*Neurography*

" reduced MCS amplitude may be present in severe cases

**Expected normal findings between attacks**

*EMG*

*Neurography*

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Drugge U, Holmberg M, Holmgren G, et al. Hereditary myopathy with lactic acidosis, succinate dehydrogenase and aconitase deficiency in northern Sweden: a genealogical study. J Med Genet 1995;32:344-347
" Haller RG, Henriksson KG, Jorfeldt L, et al. Deficiency of skeletal muscle succinate dehydrogenase and aconitase: pathophysiology of exercize in a novel human muscle oxidative defect. J Clin Invest 1991;88:1197-1206
" Linderholm H, Essen-Gustavsson B, Thornell L-E. Low succinate dehydrogenase (SDH) activity in a patient with a hereditary myopathy with paroxysmal myoglobinuria. J Intern Med 1990;228:43-52

**Modified**

" 1.4.1997, 3.4.97 ES

## FATAL INFANTILE MYOPATHY WITH CYTOCHROME C OXIDASE DEFICIENCY (COMPLEX IV DEFICIENCY)

**Etiology**

" occurs as sporadic and autosomal recessive forms
" genetic defect nor known
" selective absence of subunits VII a, b of cytochrome C oxidase

**Clinical features**

" normal at birth
" hypotonia and weakness develops during first weeks
" nearly all patients have renal tubular defect leading to aminoaciduria
" death by 1 year

**Strategy**

" demonstrate myopathic abnormalities

**Expected abnormal findings**

*EMG*

" myopathic abnormalities

*Neurography*

" MCS may show reduced amplitudes

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Bakker HD, van den Bogert C, Drewes JG, et al. Progressive generalized brain atrophy and infantile spasms associated with cytochrome c oxidase deficiency. J Inherit Metab Dis 1996;19:153-156
" DiMauro S, Mendell JR, Sahenk Z, et al. Fatal infantile mitochondrial myopathy and renal dysfunction due to cytochrome-c-oxidase deficiency. Neurology 1980;30:795-804
" Telerman-Toppet N, Biarent D, Bouton J-M, et al. Fatal cytochrome c oxidase deficient myopathy of infancy associated with mt DNA depletion. Differential involvement of skeletal muscle and cultured fibroblasts. J Inherit Metab Dis 1992;15:323-326

**Modified**

" 1.4.1997, 3.4.97 ES

## BENIGN INFANTILE MYOPATHY WITH CYTOCHROME C OXIDASE DEFICIENCY (COMPLEX IV DEFICIENCY)

**Etiology**
" sporadic
" genetic defect nor known
" selective absence of subunits VII a, b and II of cytochrome C oxidase

**Clinical features**
" normal at birth
" hypotonia and weakness develops during first weeks
" in spite of severe weakness often requiring ventilatory assistance the children recover and become normal by 2-3 years of age.
" CK is elevated, lactic acidosis

**Strategy**
" demonstrate myopathic abnormalities

**Expected abnormal findings**
*EMG*
" myopathic abnormalities
*Neurography*
" MCS may show reduced amplitudes

**Expected normal findings**
*Neurography*
" SCS

**Procedure**
*EMG*
" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**
" DiMauro S, Nicholson JF, Hays AP, et al. Benign infantile mitochondrial myopathy due to reversible cytochrome c oxidase deficiency. Ann Neurol 1983;14:226-234
" Salo MK, Rapola J, Somer H, et al. Reversible mitochondrial myopathy with cytochrome c oxidase deficiency. Arch Dis Child 1992;67:1033-1035

**Modified**
" 1.4.1997, 3.4.97 ES

## LEIGH'S SYNDROME

**Etiology**
" selective absence of all subunits of cytochrome c oxidase
" genetically heterogeneous
" autosomal recessive inheritance, X-linked inheritance and mitochondrial inheritance have been described

**Clinical features**
" onset usually during first year of life, sometimes in the neonatal period, rarely in adults
" first stage between 8-12 months is mostly with vomiting and failure to thrive
" second stage in infancy by motor regression, eye signs and altered breathing
" third stage between 2-10 years is of hypotonia and dysphagia.
" some patients present with dystonia
" hypotonia
" episodic vomiting and feeding problems
" spasticity
" hearing and visual loss
" loss of motor and verbal skills
" no muscular weakness although there is absence of all subunits of cytochrome c oxidase
" muscle biopsy can be used for the diagnosis

**Strategy**
" demonstrate normal EMG and neurography findings
" EEG shows abnormal findings

**Expected normal findings**
*EMG*
*Neurography*

**Procedure**
*EMG*
" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side
*EEG*

**References**

" Arai Y, Miyasato Y, Koide H. Characteristic changes on brain CT in a case of Leigh encephalopathy with deficiency of pyruvate dehydrogenase. Brain Dev 1991;13:457-458
" Ciafaloni E, Santorelli FM, Shanske S, et al. Maternally inherited Leigh syndrome. J Pediatr 1993;122:419-422
" Koga Y, Nonaka I, Nakao M, et al. Progressive cytochrome c oxidase deficiency in a case of Leigh's encephalomyelopathy. J Neurol Sci 1990;95:63-76
" Lera G, Bhatia K, Marsden CD. Dystonia as the major manifestation of Leigh's syndrome. Movement Dis 1994;9:642-649
" Reichmann H, Scheel H, Bier B, et al. Cytochrome c oxidase deficiency and long-chain Acyl coenzyme A dehydrogenase deficiency with Leigh's subacute necrotizing encephalopathy. Ann Neurol 1992;31:107-109
" Rahman S, Blok RB, Dahl H-HM, Danks DM, Kirby DM, Chow CW, Christodoulou J, Thorburn DR. Leigh syndrome: clinical features and biochemical and DNA abnormalities. Ann Neurol 1996;39:343-351


**Modified**

" 1.4.1997, 3.4.97 ES

## KEARNS-SAYRE SYNDROME

**Etiology**

" mitochondrial DNA shows deletions, usually there is a mixture of abnormal DNA with deletions and normal wild type DNA, the ratio determines the severity
" sporadic, very rarely transmitted

**Clinical features**

" onset before the age of 20
" progressive external ophtalmoplegia, ptosis
" myopathy
" pigmentary degeneration of the retina
" heart block
" ataxia
" hearing loss
" dementia
" peripheral neuropathy
" elevated spinal fluid proteins
" hypothyroidism

**Strategy**

" demonstrate myopathic abnormalities with proximal preponderance
" demonstrated sensory motor polyneuropathy

**Expected abnormal findings**

*EMG*

" mild myopathic abnormalities in muscles

*Neurography*

" MCS shows normal or slightly reduced CV, abnormal F-waves
" SCS shows reduced amplitudes and slight reduction of CV

*EEG*

" nonspecific generalized slowing

**Expected normal findings**

*EMG*

" in mild forms the EMG may be normal

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

*EEG*

**References**


" Degoul F, Nelson I, Lestienne P, et al. Deletions of mitochondrial DNA in Kearns-Sayre syndrome and ocular myopathies: genetic, biochemical and morphological studies. J Neurol Sci 1991,101:168-177
" Fischel-Ghodsian N, Bohlman MC, Prezant TR, et al. Deletion in blood mitochondrial DNA in Kearns-Sayre syndrome. Pediatr Res 1992,31:557-560Inui K, Fukushima H, Tsukamoto H, et al. Mitochondrial encephalomyopathies with the mutation of the mitochondrial tRNA Leu (UUR) gene. J Pediatr 1992,120:62-66
" Lestienne P, Ponsot G. Kearns-Sayre syndrome with muscle mitochondrial DNA deletion. Lancet 1988,I:885
" Shanske S, Moraes CT, Lombes A, et al. Widespread tissue distribution of mitochondrial DNA deletions in Kearns-Sayre syndrome. Neurology 1990,40:24-28
" Tulinius MH, Holme E, Kristiansson B, et al. Mitochondrial encephalomyopathies in childhood. II. Clinical manifestations and syndromes. J Pediatr 1991,119:251-259
" Zeviani M, Moraes CT, DiMauro S, et al. Deletions of mitochondrial DNA in Kearns-Sayre syndrome. Neurology 1988,38:1339-1346


**Modified**

" 30.3.1997, 3.4.97 ES

## FAMILIAL PROGRESSIVE EXTERNAL OPHTALMOPLEGIA (PEO) SYNDROME

**Etiology**

" genetically heterogeneous
" maternal inheritance, autosomal dominant inheritance and autosomal recessive inheritance has been described

" in the form with maternal inheritance mitochondrial DNA deletions have not been found
" in the dominant for multiple deletions are found

**Clinical features**

" progressive external opthalmoplegia, ptosis
" proximal muscle weakness
" in the maternally inherited form no systemic effects like heart block or CNS symptoms like in Kearns-Sayre
" in the dominantly inherited form there is ataxia, hearing loss and peripheral sensory motor polyneuropathy (the dominant form may be clinically similar to Kearns-Sayre
" CK is normal or slightly elevated, lactate and pyruvate may be elevated

**Strategy**

" demonstrate myopathic abnormalities with proximal preponderance
" in the dominant form there is also sensory motor polyneuropathy

**Expected abnormal findings**

*EMG*

" mild myopathic abnormalities in muscles, particularly in facial muscles

*Neurography*

" signs of axonal sensory motor polyneuropathy seen in dominant for of PEO

**Expected normal findings**

*EMG*

" in mild forms the EMG may be normal

*Neurography*

" in the maternally inherited form normal findings

**Procedure**

*EMG*

" m.frontalis
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Fassati A, Bordoni A, Amboni P, Fortunato F, Fagiolari G, et al. Chronic progressive external ophthalmoplegia: a correlative study of quantitative molecular data and histochemical and biochemical profile. J Neurol Sci 1994;123:140-6
" Harding AE, Petty RK, Morgan-Hughes JA. Mitochondrial myopathy: a genetic study of 71 cases. J Med Genet 1988;25:528-535
" Rowland LP, Blake DM, Hirano M, et al. Clinical syndromes associated with ragged-red fibers. Rev Neurol 1991;147:467-473

**Modified**

" 1.4.1997, 3.4.97 ES

## LATE ONSET MITOCHONDRIAL MYOPATHY

**Etiology**

" focal accumulation of deleted mitochondrial DNAin and altered energy status in muscles
" the mitochondrial alterations may due to exaggarated form of changes seen with normal aging in mitochondria
" histology shows ragged red fibers, fibers negative for cytochrome c oxidase (COX)

**Clinical features**

" onset after 65 years of age
" proximal muscle weakness and fatiguability
" ptosis in some patients

**Strategy**

" demonstrate mild myopathic changes in muscles

**Expected abnormal findings**

*EMG*

" mild myopathic changes, especially in proximal muscles

**Expected normal findings**

*EMG*

" in some described patients EMG has been normal

*Neurography*

" MCS
" SCS

**Procedure**

*Neurography*

" MCS: n.medianus/n.ulnaris and n.peroneus/tibialis on one side
" SCS: n.radialis and n.suralis on one side.

*EMG*

" one distal and proximal muscle in the lower extremities
" one distal and proximal muscle in the upper extremities
" paravertebral muscle in the thoracic region

**References**

" Johnston W, Karpati G, Carpenter S, Arnold D. Shoubridge EA. Late-onset mitochondrial myopathy. Ann Neurol 1993; 37:16-23

**Modified**

" 2.4.1997, 3.4.97 ES Björn, var kommer Torbergsens avhandling in, maternell mitokondriesjukdom med multiorgan-defekter

1. Torbergsen, T., Stålberg, E., and Bless, J. Nerve-muscle involvement in a large family with mitochondrial cytopathy. *Muscle Nerve* 14:35-41, 1991.

Var kommer Marinesco Sjögren in?

1. Torbergsen, T., Stålberg, E., Aasly, J., and Lindal, S. Myopathy in Marinesco-Sjögren syndrom: an electrophysiological study. *Acta Neurol Scand* 84:132-138, 1991.

"

## 4.7 METABOLIC MYOPATHIES

### ACID MALTASE DEFICIENCY (POMPE'S DISEASE, GLYCOGENOSIS TYPE 2)

**Etiology**
- autosomal recessive inheritance
- gene localized to chromosome 17q21-23
- at least six different mutations have been described
- three clinical forms: infantile, childhood and adult

**Clinical features**
- *infantile:* floppy infants, progressive muscle weakness, heart, liver and CNS involved, death usually before age of 2
- *childhood*: delayed motor milestones and progressive proximal muscle weakness
- *adult*: progressive proximal weakness, resembles limb-girdle dystrophy
- specific diagnosis depends on demonstration of acid maltase deficiency in fibroblasts or lymphocytes

**Strategy**
- demonstrate myopathic abnormalities , especially in proximal muscles

**Expected abnormal findings**

*EMG*
- EMG shows fibrillations and positive sharp waves, especially in paravertebral muscles
- MUP analysis shows typical myopathic abnormalities

*Neurography*
- MCS may show reduced amplitude

**Expected normal findings**

*Neurography*
- SCS

**Procedure**

*EMG*
- m.deltoideus/m.biceps brachii
- m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
- m.vastus lateralis/m.vastus medialis/m.tensor facia latae
- m.tibialis anterior

*Neurography*
- SCS: n.suralis and n.radialis on one side
- MCS: n.peroneus and n.medianus on one side

**References**


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- Boerkoel CF, Exelbert R, Nicastri C, et al. Leaky splicing mutation in the acid maltase gene is associated with delayed onset of glycogenosis type II. Am J Hum Genet 1995;56:887-897
- Lenard H, Schaub J, Keutel J, Osang M. Electromyography in type ii glycogenesis. Neuropediatrics 1974;5:410-424
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- McComas CE, Schochet SS, Morris HH, et al. The constellation of adult acid maltase deficiency--clinical, electrophysiologic, and morphologic features. Clin Neuropathol 1983;2:182-187
- Temple JK, Dunn DW, Blitzer MG, Shapira E. The 'muscular variant' of Pompe disease: clinical, biochemical and histologic characteristics. Am J Med Genet 1985;21:597-604
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**Modified**
- 1.4.1997 BF, 3.4.97 ES

### DEBRANCHING ENZYME DEFICIENCY (GLYCOGENOSIS TYPE 3)

**Etiology**
- autosomal recessive inheritance
- gene has been cloned and is localized to chromosome 1
- defect of either debranching enzyme transferase or debranching enzyme glucosidases (either one or both)

**Clinical features**
- may present in childhood or as adults
- liver is often affected, hepatomegaly

" in adults proximal and sometimes also distal weakness, in children hypotonia
" stiffness and cramping in exercising and myoglobinuria are nor common
" CK elevated 2-20 times normal
" final diagnosis is based on biochemical assay of muscle tissue

**Strategy**

" demonstrate myopathic abnormalities in muscles

**Expected abnormal findings**

*EMG*

" weak muscles show abundant fibrillations, complex repetitive discharges
" MUP analysis: typical myopathic abnormalities

*Neurography*

" in some patients mild abnormalities

**Expected normal findings**

*Neurography*

" most patients show normal findings

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Cornelis F, Bresobis N, Singer PA, et al. Clinical variations of neuromuscular disease in Debrancher deficiency. Arch Neurol 1984;41:1027-1032
" DiMauro S, Hartwig GB, Hays A, et al. Debrancher deficiency: neuromuscular disorder in 5 adults. Ann Neurol 1976;5:422-436
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" Yang-Feng TL, Zheng K, Yu J, et al. Assignment of the human glycogen debrancher gene to chromosome 1p21. Genomics 1992;13:931-934

**Modified**

" 1.4.1997, 3.4.97 ES

## BRANCHING ENZYME DEFICIENCY (GLYCOGENOSIS TYPE 4)

**Etiology**

" autosomal recessive inheritance
" deficiency of branching enzyme

**Clinical features**

" onset in infancy with failure to thrive and liver failure
" later hypotonia and muscle weakness
" death occurs in second year
" definite diagnosis is based on demonstration of deficiency of branching enzyme in fibroblasts

**Strategy**

"

**Expected abnormal findings**

*EMG*

" EMG findings have nor been described in the literature

*Neurography*

" not known

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Reusche E, Aksu F, Goebel HH, et al. A mild juvenile variant of type IV glycogenosis. Brain Dev 1992;14:36-43
" Schroder JM, May R, Shin YS, Sigmund M, Nase-Huppmeier S. Juvenile hereditary polyglucosan body disease with complete branching enzyme deficiency (type IV glycogenosis). Acta Neuropathol 1993;85:419-30

**Modified**

" 1.4.1997, 3.4.97 ES

## MYOPHOSPHORYLASE DEFICIENCY (MCARDLE'S DISEASE, GLYCOGENOSIS TYPE 5))

**Etiology**

" usually autosomal recessive inheritance, sometimes autosomal dominant inheritance
" gene location: 11q13
" myophosphorylase deficiency

**Clinical features**

" exercise intolerance and easy fatiguability

" stiffness and cramping in exercising
" myalgia
" myoglobinuria
" mild proximal weakness in one third of the patients, increases with age
" can clinically variable from very severe to very mild

**Strategy**

" demonstrate myopathic abnormalities
" exercise induced painful cramps are electrically silent unlike common cramps
" high frequency repetitive stimulation shows decrement
" definite diagnosis is based on histochemical demonstration of lack of myophosphorylase or DNA analysis

**Expected abnormal findings**

*EMG*

" the EMG may show fibrillations and positive sharp waves
" MUP analysis shows typical myopathic abnormalities
" exercise induced cramps are electrically silent

*Neurography*

" MCS may show reduced amplitude

*Decrement*

" hypothenar muscles with 20 Hz stimulation for 50 sec shows more than 25 % decrement

**Expected normal findings**

*Neurography*

" normal SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

*Decrement*

" hypothenar muscles with 20 Hz stimulation for 50 sec

*exercise induced cramp*

" electrical silence

**References**


" Chui LA, Munsat TL. Dominant inheritance of McArdle syndrome. Arch Neurol 1976;33:636-41
" Beynon RJ, Bartram C, Hopkins P, Toescu V, Gibson H, Phoenix J, Edwards RH. McArdle's disease: molecular genetics and metabolic consequences of the phenotype. Muscle Nerve 1995;Suppl 3:S18-22
" Chiado-Piat L, Mongini T, Doriguzzi C, Maniscalco M, Palmucci L. Clinical spectrum of McArdle disease: three cases with unusual expression. Eur Neurol 1993;33:208-211
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" Kristjansson K, Tsujino S, DiMauro S. Myophosphorylase deficiency: an unusually severe form with myoglobinuria. J Pediatr 1994;125:409-410
" Pourmand R, Sanders DB, Corwin HMR. Late-onset MacArdle's disease with unusual electromyographic findings. Arch Neurol 1983;40:374-377
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" Tsujino S, Shanske S, DiMauro S. Molecular genetic heterogeneity of myophosphorylase deficiency (McArdle's disease). New Engl J Med 1993;329:241-245
" Tsujino S, Shanske S, Nonaka I, DiMauro S. The molecular genetic basis of myophosphorylase deficiency (McArdle's disease). Muscle Nerve 1995;Suppl 3:S23-27


**Modified**

" 1.4.1997, 3.4.97 ES

## PHOSPHOFRUCTOKINASE DEFICIENCY (TARUI'S DISEASE, GLYCOGENOSIS TYPE 7)

**Etiology**

" may be genetically heterogeneous
" autosomal recessive inheritance
" gene location: 1cen32 and 12q have been described
" lack of phosphofructokinase
" male preponderance, male: female ratio 9:1 is not understood

**Clinical features**

" onset in childhood
" exercise intolerance and easy fatiguability
" stiffness and cramping in exercising
" myalgia
" myoglobinuria
" episodes of hemolysis and jaundice may occur, often mild hemolytic anemia
" can clinically be very variable from very severe to very mild
" CK normal or mildly elevated

**Strategy**

" demonstrate myopathic abnormalities
" high frequency repetitive stimulation shows decrement
" definite diagnosis is based on histochemical demonstration of lack of phosphofructokinase

**Expected abnormal findings**

*EMG*

" the EMG may show fibrillations and positive sharp waves
" MUP analysis shows typical myopathic abnormalities
" exercise induced cramps are electrically silent

*Neurography*

" MCS may show reduced amplitude

*Decrement*

" hypothenar muscles with 20 Hz stimulation for 50 sec shows more than 25 % decrement

**Expected normal findings**

*Neurography*

" normal SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Nakajima H, Hamaguchi T, Yamasaki T, Tarui S. Phosphofruktokinase deficiency: recent advances in molecular biology. Muscle Nerve 1995;18 suppl 3:S28-34
" Howard TD, Akots G, Bowden DW. Physical and genetic mapping of the muscle phosphofructokinase gene (PFKM): ressignment to human chromosome 12q. Genomics 1996;34:122-127
" Tarui S, Okuno G, Ikura Y, et al. Phosphofruktokinase deficiency in skeletal muscle, a new type of glyconenosis. Biochem iophys Res 1965;19:517-523

**Modified**

" 1.4.1997, 3.4.97 ES

## PHOSPHOGLYCERATE KINASE DEFICIENCY (GLYCOGENOSIS TYPE 9)

**Etiology**

" X-linked recessive inheritance
" gene location 1q13
" phosphoglycerate kinase deficiency

**Clinical features**

" most patients have CNS symptoms, mental retardation and seizures
" some may have a purely myopathic syndrome with progressive proximal muscle weakness and episodes of myoglobinuria
" exercise intolerance, easy fatiguability
" CK elevated
" forearm lactate test - no elevation of lactate

**Strategy**

" EMG is usually normal

**Expected normal findings**

*EMG*

*Neurography*

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Schroder JM, Dodel R, Weis J, et al. Mitochondrial changes in muscle phosphoglycerate kinase deficiency. Clin Neuropathol 1996;15:34-40
" Tonin P, Shanske S, Miranda AF, et al. Phosphoglycerate kinase deficiency: Biochemical and molecular genetic studies in a new myopathic variant (PGK Alberta). Neurology 1993;43:387-391
" Tsujino S, Shanske S, DiMauro S. Molecular genetic heterogeneity of phosphoglycerate kinase (PGK) deficiency. Muscle Nerve 1995;Suppl 3:S45-49

**Modified**

" 1.4.1997, 3.4.97 ES

## PHOSPHOGLYCERATE MUTASE DEFICIENCY (GLYCOGENOSIS TYPE 10)

**Etiology**

" autosomal recessive inheritance
" gene location 7p12-p13
" phosphoglycerate mutase deficiency

**Clinical features**

" most patients have CNS symptoms, mental retardation and seizures
" some may have a purely myopathic syndrome with progressive proximal muscle weakness and episodes of myoglobinuria
" exercise intolerance, easy fatiguability
" CK elevated
" forearm lactate test - subnormal elevation of lactate

**Strategy**
" demonstrate normal EMG between attacks
**Expected normal findings**
*EMG*
*Neurography*
**Procedure**
*EMG*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side
**References**
" Bresolin N, Ro Y, Reyers M, et al. Muscle phosphoglycerate mutase (PGAM) deficiency. Neurology 1983;33:1049-53
" Tsujino S, Shanske S, Sakoda S, Toscano A, DiMauro S. Molecular genetic studies in muscle phosphoglycerate mutase (PGAM-M) deficiency. Muscle Nerve 1995;Suppl 3:S50-53
**Modified**
" 1.4.1997, 3.4.97 ES

## LACTATE DEHYDROGENASE DEFICIENCY (GLYCOGENOSIS TYPE 11)

**Etiology**
" autosomal recessive inheritance
" localized to chromosome 11p15.4
**Clinical features**
" excessive fatigue
" exercise intolerance
" myalgia
" myoglobinuria
" normal muscle strength
" often skin rash
" forearm exercise test does not increase lactate
**Strategy**
" demonstrate normal EMG findings between attacks
**Expected normal findings**
*EMG*
*Neurography*
**Procedure**
*EMG*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side
**References**
" Bryan W, Lewis SF, Gunder M, et al. Muscle lactate dehyrogenase deficiency: a disorder of anaerobic glycogenolysis associated with exertional myoglobinuria. Neurology 1990;40 Supp 1:203
" Kanno T, Maekawa M. Lactate dehydrogenase M-subunit deficiencies: clinical features, metabolic background, and genetic heterogeneities. Muscle Nerve 1995;Suppl 3:S54-60
" Niebroj-Dobosz I, Fidzianska-Dolot A, Hausmanowa-Petrusewicz I. An unusual case of cardiac and skeletal muscle involvement and lactate dehydrogenase subunit-H deficiency. Acta Cardiomiologica 1994;6:5-16
" Tsujino S, Shanske S, Brownell AKW, et al. Molecular genetic studies of muscle lactate dehydrogenase deficiency in white patients. Ann Neurol 1994;36:661-665
**Modified**
" 1.4.1997 BF, 3.4.97 ES

## CARNITINE DEFICIENCY

**Etiology**
" autosomal recessive inheritance or acquired in the secondary form
" carnitine deficiency
**Clinical features**
" *myopathic form:* progressive, painless proximal muscle weakness in childhood or early adult life (infantile form has been described)
" *systemic form*: presents in early childhood with progressive cardiomyopathy and attacks resembling Reye's syndrome. Proximal muscle weakness. Death often occurs during the attacks before the age of 30
" *secondary form*: may be caused by renal failure, pregnancy, malnutrition, valproate therapy, liver failure, Reye's syndrome, myopathies, mitochodrial disorders
" CK normal in 50, others elevated
**Strategy**
" demonstrate myopathic abnormalities
**Expected abnormal findings**

*EMG*
" some fibrillations and positive sharp waves may occur
" MUP analysis shows myopathic abnormalities
*Neurography*
" MSC may show reduced amplitude

**Expected normal findings**

"
*Neurography*
" SCS
" MCS

**Procedure**

*EMG*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Breningstall GN. Carnitine deficiency syndromes. Pediatr Neurol 1990;6:75-81
" Boudin G, Mikel J, Guilcard A, et al. Fatal systemic carnitine deficiency with lipid storage in skeletal muscle, heart, liver and kidney. J Neurol Sci 1976;30:313-25
" Garavaglia B, Uziel G, Dworzak F, et al. Primary carnitine deficiency: Heterozygote and intrafamilial phenotypic variation. Neurology 1991;41:1691-1693
" Shapira Y, Glick B, Harel S, et al. Infantile idiopathic myopathic carnitine deficiency: treatment with L-carnitine. Pediatr Neurol 1993;9:35-38

**Modified**

" 1.4.1997, 3.4.97 ES

## CARNITINE PALMITYL TRANSFERASE DEFICIENCY

**Etiology**

" autosomal recessive inheritance
" CPT gene is localized to chromosome 11p11-p13
" deficiency of carnitine palmitoyl transferase
" male:female ratio 5:1
" autosomal dominant inheritance has been described

**Clinical features**

" episodes of myalgia, cramps and myalgia following prolonged exercise (>40 min) or fasting.
" between attacks muscle weakness is uncommon
" episodes can be triggered by general anesthesia, cold, emotional stress, low intake of carbohydrates, lack of sleep and high intake of lipids
" most common cause for myoglobinuria
" diagnosis is based on biochemical analysis of muscle tissue

**Strategy**

" EMG is usually normal between episodes

**Expected normal findings between attacks**

*EMG*
*Neurography*

**Procedure**

*EMG*
" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior
*Neurography*
" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Bonnefont JP, Haas R, Wolff J, et al. Deficiency of carnitine palmitoyltransferase I. J Child Neurol 1989;4:197-202
" DiMauro S, Melis di Mauro PM. Muscle carnitine palmityltransferase deficiency and myoglobinuria. Science 1973;182:929-931
" Handig I, Dams E, Taroni F, et al. Inheritance of the S113L mutation within an inbred family with carnitine palmitoyltransferase enzyme deficiency. Hum Genet 1996;97:291-293
" Mongini T, Doriguzzi C, Palmucci L, et al. Myoglobinuria and carnitine palmityl transferase deficiency in father and son. J Neurol 1991;238:323-324
" Verderio E, Cavadini P, Montermini L, et al. Carnitine palmitoyltransferase II deficiency: structure of the gene and characterization of two novel disease-causing mutations. Hum Molec Genet 1995;4:19-29

**Modified**

" 1.4.1997, 3.4.97 ES

## MYADENYLATE DEAMINASE DEFICIENCY

**Etiology**

" the clinical significance of myadenylate deaminase deficiency is not clear, it is a benign state, symptom free athletes with myadenylate deaminase deficiency have been described

" myadenylate deaminase deficiency
" autosomal recessive inheritance

**Clinical features**

" exercise intolerance
" myalgia
" symptoms develop during second to fourth decades
" CK may be mildly elevated
" diagnosis is based on histochemical analysis of muscle biopsy specimens

**Strategy**

" exclude other neuromuscular disorders
" normal findings or mild non-specific EMG abnormalities

**Expected abnormal findings**

*EMG*

" mild non-specific abnormalities

**Expected normal findings**

*EMG*

*Neurography*

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

" Baumeister FA, Gross M, Wagner DR, Pongrantz D, Eife R. Myadenylate deficiency with severe rabdomyolysis. E J Pediatr 1993;152:513-518
" Sabina RL, Swain JL, Olanow W. Myoadenylate deaminase deficiency: Functional and metabolic abnormalities associated with disruption of the perineal nucleotide cycle. J Clin Invest 1984;73:720-732
" Sabina RL, Morisaki T, Clarke P. Characterization of the human and rat myoadenylated deaminase genes. J Biol Chem 1990;265:9423-29

**Modified**

" 2.4.1997 BF, 3.4.97 ES

## 4.8 PRIMARY PERIODIC PARALYSIS

### HYPOKALEMIC PERIODIC PARALYSIS

**Etiology**

" autosomal dominant inheritance
" gene location: chromosome 1q31-q32
" mutation of the calcium channel = dihydropyridine (DHP)-receptor alpha 1 subunit

**Clinical features**

" first attacks within the second decade, invariably before the age of 30, more than half present before age of 16 years
" attacks last usually more than two hours often more than 24 hours
" attacks are precipitated by carbohydrate ingestion or strenuous exercise
" during attacks serum potassium is decreased
" CK normal between attacks and after attacks mildly elevated

**Strategy**

" during attack demonstrate reduced number of motor units and myopathic abnormalities
" between attacks patients with fixed weakness show myopathic abnormalities on EMG
" demonstrate abnormal exercise test between attacks

**Expected abnormal findings during attack**

*Neurography*

" MCS: reduced amplitude of the M wave

*EMG*

" insertional activity is reduced
" reduced interference pattern
" reduction in duration and amplitude of MUPs

*Decrement studies*

" facilitation of responses up to 400 %

**Expected normal findings during attack**

" SCS

**Expected abnormal findings between attacks**

*EMG*

" in patients with permanent weakness reduction in duration and amplitude of MUPs

*Exercise test*

" abnormal initial increase in M amplitude and successively abnormal decrease

**Expected normal findings between attacks**

*EMG*

" in patients without permanent weakness normal EMG

*Neurography*

" SCS
" MCS

**Procedure**

*Neurography*

" MCS: n.medianus and n.peroneus unilaterally

*EMG*

" m.deltoideus
" m.interosseus dorsalis/m.opponens pollicis
" m.vastus lateralis
" m.tibialis anterior

*Decrement (during attacks)*

" hypothenar muscles

*Exercise test (between attacks)*

" McManis PG, Lambert EH, Daube JR. Exercise test in periodic paralysis. Muscle Nerve 1986,9:704
" hypothenar muscles: M wave amplitude: for 2-5 minute maximal voluntary exercise 15 sec and 5 sec rests
" in controls M amplitude increases immediately after exercise less than 27 % and decreases less than 30 % 5 min after the exercise test
" in periodic paralysis increase after exercise >35% and successive drop in amplitude >27%

**Note**

" specific diagnosis can be obtained with genetic molecular methods
" muscle biopsy shows distinctive vacuoles
" provocative tests: glucose loading can be used

**References**

" Boerman RH, Ophoff RA, Links TP, et al. Mutation in DPH receptor alpha 1 subunit (CACLN1A3) gene in a Dutch family with hypokalemic periodic paralysis. J Med Genet 1995;32:44-47
" Fontaine B, Vale-Santos J, Jurkat-Rott K, Reboul J, et al. Mapping of the hypokalaemic periodic paralysis (HypoPP) locus to chromosome 1q31-32 in three European families. Nature Genetics 1994;6:267-272
" Links TP, van-der-Hoeven JH, Zwarts MJ. Surface EMG and muscle fibre conduction during attacks of hypokalaemic periodic paralysis. J Neurolo Neurosurg Psychiatry 1994;57:632-634
" Kantola IM, Tarssanen LT. Familial hypokalaemic periodic paralysis. J Neurolo Neurosurg Psychiatry 1992;55:322-324
" McManis PG, Lambert EH, Daube JR. Exercise test in periodic paralysis. Muscle Nerve 1986,9:704-708

**Modified**

" 1.4.1997, 3.4.97 ES

## HYPERKALEMIC PERIODIC PARALYSIS

**Etiology**

" autosomal dominant inheritance
" gene location chromosome 17q13,1
" mutation of muscle alpha-subunit of the sodium channel
" this disorder is more heterogeneous than hypokalemic periodic paralysis
" hyperkalemic periodic paralysis is allelic to paramyotonia congenita and myotonia fluctuans

**Clinical features**

" first attacks during the first decade
" initially attacks are rare but they become more frequent
" duration of attacks 15 min to 4 hours
" cold, emotional stress, glucocorticoids and pregnancy aggravate attacks
" after strenuous exercise weakness can follow within minutes, sustained exercise may prevent weakness
" attacks precipitated by fasting or exercise
" the serum potassium level reaches 5-6 mmol/l and rarely reaches cardiotoxic levels

**Strategy**

" during attack demonstrate reduced number of motor units and myopathic abnormalities
" between attacks patients with fixed weakness show myopathic abnormalities on EMG
" demonstrate abnormal exercise test between attacks

**Expected abnormal findings during attack**

*Neurography*

" MCS: reduced amplitude of the M wave

*EMG*

" reduced interference pattern
" reduction in duration and amplitude of MUPs
" myotonic discharges tend to diminish during attacks

*Decrement studies*

" facilitation of responses up to 400 %

**Expected normal findings during attack**

" SCS

**Expected abnormal findings between attacks**

*EMG*

" in patients with permanent weakness reduction in duration and amplitude of MUPs
" myotonic discharges

*Exercise test*

" abnormal initial increase in M amplitude and successively abnormal decrease

**Expected normal findings between attacks**

*EMG*

" in patients without permanent weakness normal EMG

*Neurography*

" SCS
" MCS

**Procedure**

*Neurography*
" MCS: n.medianus and n.peroneus unilaterally
*EMG*
" m.deltoideus
" m.interosseus dorsalis/m.opponens pollicis
" m.vastus lateralis
" m.tibialis anterior
*Decrement (during attacks)*
" hypothenar muscles
*Exercise test (between attacks)*
" McManis PG, Lambert EH, Daube JR. Exercise test in periodic paralysis. Muscle Nerve 1986,9:704
" hypothenar muscles: M wave amplitude: for 2-5 minute maximal voluntary exercise 15 sec and 5 sec rests
" in controls M amplitude increases immediately after exercise less than 27 % and decreases less than 30 % 5 min after the exercise test
" in periodic paralysis increase after exercise >35% and successive drop in amplitude >27%

**Note**
" specific diagnosis can be obtained with genetic molecular methods
" hyperkalemic periodic paralysis can be differentiated from paramyotonia congenita using cold provocation: in paramyotonia congenita cooling of the muscle to 20 C eliminates myotonic discharges and voluntary activity
" muscle biopsy shows vacuoles that are smaller than in hypokalemic periodic paralysis
" provocative tests: potassium loading can be used

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**Modified**
" 1.4.1997 BF, 3.4.97 ES

## 4.9 MISCELLANEOUS MYOPATHIES

### BENT SPINE SYNDROME (ISOLATED NECK EXTENSOR MYOPATHY)

**Etiology**
" heterogenous
" some hereditary forms have been described

**Clinical features**
" onset in old age most described patients are above 65 years of age
" weakness of paraspinal muscles on standing, resulting in a bent spine or inability to maintain the head erect (dropped head syndrome)

**Strategy**
" demonstrate myopathy in the paraspinal muscles, especially in the cervical and thoracic region
" differentiate from ALS, more widespread myopathies, especially polymyositis

**Expected abnormal findings**
*EMG*
" active myopathic findings in paraspinal muscles

**Expected normal findings**
*EMG*
" limb muscles
*Neurography*
" MCS
" SCS

**Procedure**
*Neurography*
" MCS n.medianus, n.ulnaris and n.peroneus on one side
" SCS: n.radialis and n.suralis on one side.
*EMG*
" Paraspinal muscles in the cervical, thoracic and lumbar region
" one distal and proximal muscle in the lower extremities.

**References**

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**Modified**
" 2.4.1997, 3.4.97 ES

### MALIGNANT HYPERTHERMIA

**Etiology**

" genetic basis is incompletely understood
" autosomal dominant inheritance is most common
" gene location: chromosome 19q13.1 in a portion of the patients
" mutation of the ryanodine receptor gene
" patients with central core disease (gene mutation on the same allele as above) has a susceptibility to malignant hyperthermia

**Clinical features**

" malignant hyperthermia is triggered by volatile anesthetic agent
" onset is often more abrupt if succinycholine is used
" once malignant hyperthermia is induced the temperature rises and may exceed 43 C
" 75 % show muscle rigidity
" may proceed to rigor and death
" serum potassium, CK, ionized calcium, myoglobin and sodium is elevated
" pH falls below 7.00
" treatment by intravenous dantrolone sodium and cooling

**Strategy**

" between attacks no EMG abnormalities

**Expected normal findings**

*Neurography*

" SCS
" MCS

*EMG*

**Procedure**

*Neurography*

" MCS: n.radialis and n.peroneus unilaterally

*EMG*

" m.deltoideus
" m.interosseus dorsalis/m.opponens pollicis
" m.vastus lateralis
" m.tibialis anterior

**References**

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**Modified**

" 1.4.1997, 3.4.97 ES

## NEUROLEPTIC MALIGNANT SYNDROME

**Etiology**

" not related with malignant hypertermia
" sporadic
" less than 1% of persons taking neuroleptics, especially haloperidol and fluphenazine develop neuroleptic malignant syndrome

**Clinical features**

" occurs especially in young men on neuroleptic drug treatment
" fever, rigidity, confusion, tachycarida
" rhabdomyolysis
" myoglobinuria

**Strategy**

" EMG is usually normal between episodes

**Expected normal findings between attacks**

*EMG*

*Neurography*

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor facia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**References**

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**Modified**

" 1.4.1997, 3.4.97 ES

## RIPPLING MUSCLE DISEASE

**Etiology**

" autosomal dominant, chromosome 1q41
" may be genetically heterogeneous

**Clinical features**

" onset of symptoms in teens
" symptoms worst at age of onset, subsequently symptoms decrease
" muscle cramps and stiffness, especially with exercise
" stiffness most marked in proximal muscles
" muscle hypertrophy
" mechanical tapping of the muscle induces local mounding and rippling muscle contractions
" CK is mildly or moderately elevated (up to 17 times normal)

**Strategy**

" demonstrate that there is no electric muscle activity related with the rippling muscle contractions

**Expected abnormal findings**

*EMG*

" mechanical tapping induces electrically silent muscle contractions

**Expected normal findings**

*EMG*

" insertional activity
" MUP analysis

*Neurography*

" SCS
" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus, n.tibialis, n.ulnaris and n.medianus on one side

**References**

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**Modified**

" 1.4.1997, 3.4.97 ES

# 5. LOCAL NERVE LESIONS

## 5.1 SHOULDER, NECK AND UPPER EXTREMETIES

### CARPAL TUNNEL SYNDROME (CTS)

**Etiology**

" chronic compression of the median nerve in the carpal tunnel

**Clinical features**

" by far the most common local nerve lesion
" paresthesias of digits 1-4, especially during the night or following use of hands
" in early cases only intermittent numbness in more severe entrapments constant numbness
" affects women more often than men (ratio 4:1)
" most patients are 40-60 years of age
" younger onset of symptoms is not unusual, occurs rarely in children
" in severe cases atrophy of the thenar muscles
" onset is insidious in most patients
" pain in the wrist and forearm, often also in the shoulder region
" predisposing factors: obesity, heavy manual work, pregnancy, wrist fractures, rheumatoid arthritis, diabetes, tenosynovitis of finger flexor tendons, hypothyreosis, amyloidosis, acromegaly

**Strategy**

" confirm local lesion of median nerve in the carpal tunnel

Assess

" severity: mild - moderate -severe - total
" pathophysiology: conduction block - demyelination -axonal degeneration

**Expected abnormal findings**

*Neurography*

" reduced median nerve SCV or prolonged sensory latency at the wrist
" in moderate and severe cases prolonged distal motor latency of the median nerve
" in moderate to severe cases reduced SCS amplitude
" in severe cases reduced MCS amplitude

*EMG*

- in moderate to severe cases m.abductor pollicis brevis show neurogenic findings

**Expected normal findings**

- SCS: ulnar and radial nerves
- MCS: ulnar nerve

**Procedure**

*Neurography (always bilaterally)*

- SCS, median nerve digit 3 and one other digit to wrist, (palm to wrist may be helpful)
- SCS, ulnar nerve digit 5 to wrist, (palm to wrist may be helpful)
- sometimes comparison of the median nerve SCS with radial nerve forearm to thumb may be helpful
- MCS median nerve

*EMG (optional: if it is necessary to do EMG to exclude a proximal lesion:)*

- m.opponens pollicis/m.abductor pollicis brevis
- m.interosseus dorsalis I
- m.flexor carpi radialis/flexor pollicis longus
- m.extensor indicis proprius
- m.triceps

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**Revised**

" BF 1.2.1997, BF 12.4.1997

## MEDIAN NERVE LESION ABOVE THE WRIST

**Etiology**

" trauma, especially with knife and glass

**Clinical features**

" loss of sensation and paresthesia of digits 1-4

" weakness and/or atrophy of the thenar muscles
" pain is variable
" trauma caused by sharp objects, especially knifes and glass may cut only some fascicles; this may case limited symptoms

**Strategy**

" confirm local lesion of median nerve above the wrist
" differentiate from carpal tunnel syndrome

Assess

" severity: mild - moderate -severe - total
" pathophysiology: conduction block - demyelination -axonal degeneration

**Expected abnormal findings**

*Neurography*

" reduced n.medianus SCV in nerve lesions caused by compression
" in moderate and severe cases prolonged distal motor latency of the median nerve
" reduced n.medianus SCS amplitude, especially in partial lesions due to cutting with sharp objects
" in moderate or severe cases reduced MCS amplitude

*EMG*

" in moderate to severe cases m.abductor pollicis brevis show neurogenic findings

**Expected normal findings**

" SCS: ulnar and radial nerves
" MCS: ulnar nerve

**Procedure**

*Neurography*

" SCS, median nerve digit 3 and one other digit to wrist crease and 50 mm above wrist crease
" SCS, ulnar nerve digit 5 to wrist
" MCS median nerve bilaterally

*EMG*

" m.opponens pollicis/m.abductor pollicis brevis

*the following muscles are optional, required only if it is necessary to do EMG to exclude a proximal lesion*

" m.interosseus dorsalis I
" m.flexor carpi radialis/flexor pollicis longus
" m.extensor indicis proprius
" m.triceps
" m.biceps
" m.deltoideus

**Revised**

" BF 1.2.1997

## ANTERIOR INTEROSSEUS NERVE LESION

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy) by far most common cause of n.interosseus anterior neuropathy
" traumatic injury in association with elbow luxation and other injuries
" often described in textbooks as an entrapment neuropathy (anterior interosseus syndrome), we have not encountered a real entrapment at this site and are doubtful as to whether such an entrapment really exists

**Clinical features**

" usually acute onset within hours
" often severe pain in the anterior part of the forearm, pain subsides after a few days or weeks
" flexion weakness of the distal phalanx of the thumb and forefinger
" no sensory loss
" prognosis with conservative treatment is usually good

**Strategy**

" demonstrate neurogenic EMG findings in muscles in muscles innervated by n.interosseus anterior
" normal findings in other muscles innervated by n.medianus

**Expected abnormal findings**

*EMG :*

" neurogenic findings in m.pronator quadratus and m.flexor pollicis longus

*Neurography*

" MCS latency to pronator quadratus prolonged

**Expected normal findings**

*EMG*

" m.flexor carpi radialis
" m.abductor pollicis brevis
" m.pronator teres

*Neurography*

" MCS: n.medianus
" SCS: n.medianus

**Procedure**

*EMG :*

" m. pronator quadratus (n.interosseus anterior, distal part)
" m.flexor pollicis longus (n.interosseus anterior, proximal part)
" m.flexor carp radialis/m.pronator teres (n.medianus, proximal part)
" m.opponens pollicis/m.abductor pollicis brevis (n.medianus, distal part)
" m. interosseus dorsalis (n.ulnaris)
" m.extensor digitorum communis (n.radialis)

*Neurography*

" MCS latency elbow pronator quadratus bilaterally
" n.medianus MCS in the forearm to m.abductor pollicis brevis
" n.medianus SCS to digit 3

*the following muscles are optional, required only if it is necessary to do EMG to exclude a proximal lesion*

" m.triceps
" m.biceps
" m.deltoideus

**Note**

" we discourage the use of the term anterior interosseus syndrome which alludes to an entrapment neuropathy

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**Revised**

" BF 1.2.1997, 12.4.1997

## MEDIAN NERVE LESION AROUND THE ELBOW

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy) by far most common cause of n.interosseus anterior neuropathy
" traumatic injury in association with elbow luxation, supracondylar humerus fractures and other injuries
" often described in textbooks as an entrapment neuropathy (pronator syndrome) in the region where the median nerve passes thorough the pronator teres muscle; we have never encountered a real entrapment at this site and are doubtful as to whether such an entrapment really exists

**Clinical features**

" paresthesia of fingers 1-4
" in moderate or severe cases weakness of thumb abduction, opposition and flexion, wrist flexion and finger flexion

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the median nerve distal to the pronator muscle
" differentiate from other peripheral nerve lesions

**Expected abnormal findings**

*EMG*

" m.flexor pollicis longus
" m.opponens pollicis/abductor pollicis brevis
" m.flexor digitorum profundus and superficialis

*Neurography*

" reduced CV and/or conduction block of median nerve in the proximal part of the forearm

**Expected normal findings**

*EMG*

" m.pronator teres and m.flexor carpi radialis ( innervated proximal to the lesion)
" muscles innervated by the ulnar and radial nerve

*Neurography*

" normal ulnar and radial nerve MCS and SCS

**Procedure**

*EMG:*

" m.flexor pollicis longus (n.interosseus anterior)
" m.flexor carp radialis/m.pronator teres (n.medianus, proximal part)
" m.abductor pollicis brevis (n.medianus, distal part)
" m.interosseus dorsalis (n.ulnaris)
" m.extensor digitorum communis (n.radialis)

*Neurography*

" MCS latency elbow pronator quadratus
" n.medianus MCS in the forearm to thenar muscles
" n.medianus SCS to digit 3

**Note**

" we discourage the use of the term pronator syndrome which alludes to an entrapment neuropathy

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**Revised**
" 6.4.1997

## MEDIAN NERVE LESION ABOVE THE ELBOW (AT STRUTHER'S LIGAMENT)

**Etiology**
" described in the literature as an entrapment neuropathy in lower part of the upper arm, the entrapment is due to a ligament from supracondylar process to the medial epicondyle. We have never encountered a real entrapment at this site and are doubtful as to whether such an entrapment really exists.

**Clinical features**
" paresthesia of fingers 1-4
" in moderate or severe cases weakness of thumb abduction, opposition and flexion, wrist flexion and finger flexion
" plain X-rays show Struther's ligament ligament from supracondylar process to the medial epicondyle

**Strategy**
" demonstrate neurogenic EMG findings in muscles innervated by the median nerve in the forearm
" differentiate from other peripheral nerve lesions.

**Expected abnormal findings**
*EMG*
" neurogenic EMG findings in muscles innervated by the median nerve.
*Neurography*
" reduced CV and/or conduction block of median nerve in distal part of the upper arm.

**Expected normal findings**
*EMG*
" muscles innervated by the n.ulnaris and n.radialis nerve.
*Neurography*
" n.ulnaris and n.radialis MCS and SCS.

**Procedure**
*EMG :*
" m.flexor pollicis longus (n.interosseus anterior)
" m.flexor carp radialis/m.pronator teres (n.medianus, proximal part)
" m.opponens pollicis/abductor pollicis brevis (n.medianus, distal part)
" m.interosseus dorsalis (n.ulnaris)
" m.extensor digitorum communis (n.radialis)
*Neurography*
" MCS latency elbow pronator quadratus
" n.medianus MCS in the forearm to thenar muscles
" n.medianus SCS to digit 3

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**Revised**

" 6.4.1997 BF, 7.5.1997 BF

## RADIAL NERVE LESION IN THE UPPER ARM

**Etiology**

" temporary compression of the radial nerve in radial groove, often during sleep following heavy drinking
" secondary to fracture of the humerus
" torniquet paralysis
" erroneous injections

**Clinical features**

" wrist drop in severe cases
" weakness of finger extension, wrist extension, and elbow flexion (may be compensated by m.biceps brachii)
" tendon reflex of m.brachioradialis reduced or absent
" no weakness of elbow extension and m.triceps tendon reflex normal
" inconsistent sensation loss over the dorsal side of the hand between metacarpal bones 1 and 2

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the radial nerve below the radial groove
" assess severity and pathophysiology (axonal lesion/conduction block) to be able to give a prognosis

**Expected abnormal findings**

*EMG*

" denervation of m.brachioradialis
" denervation of wrist extensors
" denervation of finger extensors

*Neurography*

" radial nerve MCS : conduction block in the radial groove (not commonly assessed)
" radial nerve MCS: reduced amplitude
" radial nerve SCS: reduced amplitude (normal if it is a pure conduction block)

**Expected normal findings**

*EMG*

" m.triceps
" muscles innervated by n.medianus and n.ulnaris

*Neurography*

" n.ulnaris and n.medianus

**Procedure**

*Neurography*

" SCS: n.radialis, n.medianus, n.ulnaris
" MCS: n.radialis (fractionated across the radial groove)(optional)

*EMG*

" m.biceps brachii
" m.triceps
" m.brachioradialis/m.extensor carpi radialis
" m.extensor digitorum communis/m.extensor indicis
" m.opponens pollicis/m.abductor pollicis brevis
" m.interosseus dorsalis

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**Revised**

" BF 1.2.1997, BF 28.4.1997

## RADIAL NERVE LESION IN THE AXILLA

**Etiology**

" compression by crutches
" trauma to the shoulder

**Clinical features**

" wrist drop in severe cases
" weakness of finger extension, wrist extension, elbow flexion and elbow extension
" m.triceps tendon reflex absent
" inconsistent sensation loss over the dorsal side of the hand between metacarpal bones 1 and 2

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the radial nerve
" assess severity and pathophysiology (axonal lesion/conduction block) to be able to give a prognosis

**Expected abnormal findings**

*EMG*
" m.triceps
" m.brachioradialis/m.extensor carpi radialis
" extensor digitorum communis
*Neurography*
" n.radialis MCS : conduction block in the axilla (not commonly assessed)
" n.radialis MCS: reduced amplitude
" n.radialis SCS: reduced amplitude (normal if it is a pure conduction block)

**Expected normal findings**
*EMG*
" m.deltoideus (axillary nerve branches from the posterior fascicle together with the radial nerve)
" n.axillaris, n.ulnaris and n.medianus innervated muscles
*Neurography*
" ulnar and median nerves

**Procedure**
*Neurography*
" SCS: n.radialis, n.medianus, n.ulnaris
" MCS: n.radialis (fractionated across the radial groove)(optional)
*EMG*
" m.deltoideus (posterior fascicle)
" m.biceps
" m.triceps
" m.brachioradialis
" m.extensor digitorum communis/extensor indicis
" m.opponens pollicis/m.abductor pollicis brevis (n.medianus)
" m.interosseus dorsalis (n.ulnaris)

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**Revised**
" BF 1.2.1997

## POSTERIOR INTEROSSEUS NERVE LESION

**Etiology**
" acute idiopathic mononeuropathy (acute neuralgic amyotrophy)
" trauma to the forearm, laceration
" Monteggia-fracture
" entrapment is extremely rare, if at all existent

**Clinical features**
" weakness of finger extension
" no weakness of wrist extension, elbow flexion or elbow extension and m.triceps tendon reflex normal
" no sensory abnormalities
" usually painless but there may be local pain 5-8 cm distal to the lateral epicondyle

**Strategy**
" demonstrate neurogenic EMG findings in muscle innervated by the deep branch of the radial nerve

**Expected abnormal findings**
*EMG*
" m.extensor digitorum communis/m.extensor indicis proprius
*Neurography*
" n.radialis MCS (elbow to forearm) abnormal (CV reduced or conduction block)

**Expected normal findings**
*EMG*
" m.extensor carpi radialis
" m. brachioradialis
*Neurography*
" SCS n.radialis

**Procedure**
*Neurography*
" SCS: n.radialis, n.medianus, n.ulnaris
" MCS: n.radialis (fractionated across the arcade of Frohse)
*EMG*
" m.triceps (n.radialis proximal to lesion)
" m.brachioradialis/m.extensor carpii radialis longus (n.radialis proximal to lesion)
" m.extensor digitorum communis/m.extensor indicis
" m.opponens pollicis/m.abductor pollicis brevis (n.medianus)
" m.interosseus dorsalis (n.ulnaris)

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**Revised**

" 12.5.1997 BF, 1.2.1997 BF

## LESION OF THE SENSORY RADIAL NERVE THE FOREARM (WARTENBERG'S SYNDROME)

**Etiology**

" temporary compression of the superficial radial nerve in the forearm by tight handcuffs or wrist watch
" surgery for de Quervain's tenosynovitis
" intravenous catethers in the radial aspect of the forearm

**Clinical features**

" numbness and/or loss of sensation over the dorsal side of the hand between metacarpal bones 1 and 2, sometimes on the dorsal side of fingers 1-3
" sometimes when the lesion is caused by a sharp cutting object only one branch of n.radialis is damaged

**Strategy**

" demonstrate lesion of the superficial radial nerve or one of its branches

**Expected abnormal findings**

*Neurography*

" SCS: n.radialis low amplitude and/or reduced CV

**Expected normal findings**

*Neurography*

" SCS: n.medianus, n.ulnaris

*EMG*

" m.extensor digitorum communis
" m.interosseus dorsalis I
" m.abductor pollicis brevis

**Procedure**

*Neurography*

" SCS: n.radialis, n.medianus, n.ulnaris
" it may be necessary to study separately the n.radialis branches to the thumb, forefinger and long finger

*EMG*

" m.extensor digitorum communis
" m.opponens pollicis/m.abductor pollicis brevis
" m.interosseus dorsalis

**References**

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**Revised**

" BF 1.2.1997, 12.4.1997

## ULNAR NERVE LESION IN THE CUBITAL TUNNEL (CUBITAL TUNNEL SYNDROME)

**Etiology**

" entrapment of the ulnar nerve at the flexor retinaculum of the m.flexor carpi ulnaris 1-2 cm distal to the medial epicondyle

**Clinical features**

" paresthesia of fingers 4 and 5
" weakness of spreading of the fingers
" in severe cases atrophy of the intrinsic hand muscles except the thenar muscles
" pain sometimes present over the elbow region, pain is not at all as prominent as in carpal tunnel syndrome
" the elbow does not usually show clinically or radiologically deformity

**Strategy**

" demonstrate nerve conduction abnormality just distal to the medial epicondyle
" demonstrate neurogenic EMG findings in muscles innervated by the ulnar nerve
" exclude proximal nerve lesions (lower part of plexus and C8 radiculopathy)

**Expected abnormal findings**

*EMG*

" m.interossseus dorsalis/m.abductor digiti minimi
" m.flexor carpi ulnaris

*Neurography*

" MCS, n.ulnaris: reduced CV and/or conduction block immediately distal to the medial epicondyle
" SCS, n.ulnaris digits 4 and 5 and n.ulnaris ramus dorsalis reduced amplitude
" ulnar nerve inching: abnormal finding 10-20 mm distal to the medial epicondyle

**Expected normal findings**

*EMG*

" m.extensor indicis proprius (C7-C8)
" m.abductor pollicis brevis (may be innervated from n.ulnaris and show abnormality)
" paravertebral muscles

*Neurography*

" SCS and MCS n.medianus and n.radialis
" SCS: n.cutaneus antebrachii medialis

**Procedure**

*Neurography*

" MCS n.ulnaris, fractionated across the elbow
" MCS, SCS n.ulnaris
" SCS, n.ulnaris ramus dorsalis
" SCS n.medianus
" inching across the elbow: n.ulnaris

*EMG*

" m.interosseus dorsalis I (distal n.ulnaris)
" m.opponens pollicis/m.abductor pollicis brevis (n.medianus)
" m.flexor carpi ulnaris (proximal n.ulnaris)
" m.extensor indicis proprius (C8 and inferior trunk)

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**Revised**

" 1.7.1997 BF, 3.4.1997 BF

## ULNAR NERVE LESION AT THE MEDIAL EPICONDYLE (ULNAR SULCUS)

**Etiology**

" entrapment in the ulnar sulcus at the medial epicondyle or just proximal to it
" temporary compression during sleep (often following alcohol consumption) or anesthesia
" trauma to the elbow

**Clinical features**

" paresthesia of fingers 4 and 5
" weakness of spreading of the fingers
" in severe cases atrophy of the intrinsic hand muscles except the thenar muscles
" pain sometimes present over the elbow region, pain is not at all as prominent as in carpal tunnel syndrome
" if the lesion is an entrapment the elbow often shows clinically or radiologically deformity (limited extension of the elbow)

**Strategy**

" demonstrate nerve conduction abnormality at the medial epicondyle
" demonstrate neurogenic EMG findings in muscles innervated by the ulnar nerve
" exclude proximal nerve lesions (lower part of plexus and C8 radiculopathy)

**Expected abnormal findings**

*EMG*

" m.interossseus dorsalis/m.abductor digiti minimi
" m.flexor carpi ulnaris

*Neurography*

" MCS, n.ulnaris: reduced CV and/or conduction block immediately distal to the medial epicondyle
" SCS, n.ulnaris digits 4 and 5 and n.ulnaris ramus dorsalis reduced amplitude
" ulnar nerve inching: abnormal finding at the medial epicondyle or just proximal to it

**Expected normal findings**

*EMG*

" m.extensor indicis proprius (C7-C8)
" m.opponens pollicis/m.abductor pollicis brevis (may be innervated from n.ulnaris and show abnormality)
" paravertebral muscles

*Neurography*

" SCS and MCS n.medianus and n.radialis
" SCS: n.cutaneus antebrachii medialis

**Procedure**

*Neurography*

" MCS n.ulnaris, fractionated across the elbow
" MCS, SCS n.ulnaris
" SCS, n.ulnaris ramus dorsalis
" SCS n.medianus
" inching across the elbow: n. ulnaris

*EMG*

" m.interosseus dorsalis I (distal n.ulnaris)
" m.abductor pollicis brevis (n.medianus)
" m.flexor carpi ulnaris (proximal n.ulnaris)
" m.extensor indicis proprius (C8 and inferior trunk)

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**Revised**
" 1.7.1997 BF, 3.4.1997 BF, 12.4.1997

## ULNAR NERVE LESION AT THE WRIST

**Etiology**
" entrapment in the canal of Guyon (usually due to a lipoma, ganglion or aneurysm ), rare
" temporary or chronic compression to the hypothenar region (due to bicycle bars, crutches)
" trauma, especially sharp cutting objects

**Clinical features**
" paresthesia and/or loss of sensation of fingers 4-5
" no alteration of the sensation over the dorsal side of the hand innervated by r.dorsalis n.ulnaris
" weakness of ulnar innervated hand muscles

**Strategy**
" demonstrate lesion of the ulnar nerve at the wrist
" assess severity and pathophysiology

**Expected abnormal findings**
*EMG*
" neurogenic EMG findings in m.interossseus dorsalis/m.abductor digiti minimi
*Neurography*
" ulnar MCS reduced amplitude and prolonged DLAT
" ulnar SCS reduced CV at the wrist and reduced AMPL

**Expected normal findings**
*EMG*
" m.flexor carpi ulnaris
" m.extensor indicis proprius
*Neurography*
" median and radial nerves
" n.ulnaris ramus dorsalis

**Procedure**
*Neurography*
" MCS n.ulnaris, fractionated across the elbow
" MCS, SCS n.ulnaris
" SCS, n.ulnaris ramus dorsalis
" SCS n.medianus
" inching across the elbow: n.ulnaris
*EMG*
" m.interosseus dorsalis I (distal n.ulnaris)
" m.abductor pollicis brevis (n.medianus)
" m.flexor carpi ulnaris (proximal n.ulnaris)
" m.extensor indicis proprius (C8 and truncus inferior)

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**Revised**
" 1.7.1997BF , 12.4.1997 BF

## DEEP MOTOR BRANCH OF THE ULNAR NERVE LESION AT THE WRIST

### Etiology
" entrapment in the canal of Guyon (usually due to a lipoma, ganglion or aneurysm )
" temporary or chronic compression to the hypothenar region (due to bicycle bars, crutches)
" trauma, especially sharp cutting objects

### Clinical features
" weakness and atrophy of intrinsic hand muscles, except thenar muscles
" no sensory abnormality

### Strategy
" demonstrate lesion of the deep motor branch of the ulnar nerve at the wrist
" assess severity and pathophysiology

### Expected abnormal findings
*EMG*
" neurogenic EMG findings in m.interossseus dorsalis/m.abductor digiti minimi
*Neurography*
" ulnar MCS reduced amplitude and prolonged DLAT

### Expected normal findings
*EMG*
" m.flexor carpi ulnaris
" m.extensor indicis proprius
*Neurography*
" SCV: n.ulnaris, n.medianus and n.radialis

### Procedure
*Neurography*
" MCS n.ulnaris, fractionated across the elbow
" MCS, SCS n.ulnaris
" SCS, n.ulnaris ramus dorsalis
" SCS n.medianus
" inching across the elbow: n.ulnaris
*EMG*
" m.interosseus dorsalis I/m.abductor digiti minimi (distal n.ulnaris)
" m.abductor pollicis brevis (n.medianus)
" m.flexor carpi ulnaris (proximal n.ulnaris)
" m.extensor indicis proprius (C8 and truncus inferior)

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### Revised
" 12.4.1997 BF, 15.4.1997 BF

## SUPERFICIAL SENSORY BRANCH OF THE ULNAR NERVE LESION AT THE WRIST

### Etiology
" entrapment in the canal of Guyon (usually due to a lipoma, ganglion or aneurysm )
" temporary or chronic compression to the hypothenar region (due to bicycle bars, crutches)
" trauma, especially sharp cutting objects

### Clinical features
" paresthesia and/or loss of sensation of fingers 4-5

**Strategy**
" demonstrate lesion of the superficial sensory branch of the ulnar nerve at the wrist
" assess severity and pathophysiology

**Expected abnormal findings**
*Neurography*
" ulnar SCS reduced CV at the wrist and amplitude

**Expected normal findings**
*EMG*
" m.abductor digiti minimi/m.interosseus dorsalis I
" m.flexor carpi ulnaris
" m.extensor indicis proprius
*Neurography*
" SCS: n.medianus and n.radialis, n.ulnaris ramus dorsalis
" MCS: n.ulnaris, n.medianus

**Procedure**
*Neurography*
" MCS n.ulnaris, fractionated across the elbow
" MCS, SCS n.ulnaris
" SCS, n.ulnaris ramus dorsalis
" SCS n.medianus
" inching across the elbow: n.ulnaris
*EMG*
" m.interosseus dorsalis I (distal n.ulnaris)
" m.abductor pollicis brevis (n.medianus)
" m.flexor carpi ulnaris (proximal n.ulnaris)
" m.extensor indicis proprius (C8 and truncus inferior)

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**Revised**
" 1.7.1997 BF, 12.4.1997 ES, 15.4.1997 BF

## LATERAL CUTANEOUS NERVE OF THE FOREARM LESION

**Etiology**
" isolated lesions rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets
" trauma in association with venipuncture of v.cephalica
" surgery in the antecubital fossa
" fractures of the elbow and proximal part of forearm

**Clinical features**
" loss of sensation over the anterior lateral part of the forearm (n.cutaneus antebrachii lateralis)

**Strategy**
" demonstrate sensory abnormality limited to n.cutaneus antebrachii lateralis

**Expected abnormal findings**
*Neurography*
" SCS: n.cutaneus antebrachii lateralis

**Expected normal findings**
*EMG*
" m.biceps brachii
" m.brachialis/m.coracobrachialis
" m.deltoideus
" m.brachioradialis
" m.triceps brachii

**Procedure**
*EMG*
" m.biceps brachii (n.musculocutaneus)
" m.brachioradialis (C6, plexus brachialis: upper trunk)
" m.flexor carpi radialis (plexus brachialis: lateral fascicle)
" m.triceps (posterior fascicle)
*Neurography*
" n.cutaneus antebrachii lateralis bilaterally
" n.radialis

**References**
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**Revised**
" 1.7.1997 BF, 23.4.1997 BF

## MEDIAL CUTANEOUS NERVE OF THE FOREARM LESION

**Etiology**
" isolated lesions rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets
" trauma in association with venipuncture of v.basilica
" surgery in the antecubital fossa
" fractures of the elbow and proximal part of forearm

**Clinical features**
" loss of sensation over the anterior medial part of the forearm (n.cutaneus antebrachii medialis)

**Strategy**
" demonstrate sensory abnormality limited to n.cutaneus antebrachii medialis
" differentiate from Th1 radiculopathy

**Expected abnormal findings**
*Neurography*
" SCS: n.cutaneus antebrachii medialis

**Expected normal findings**
*EMG*
" m.abductor pollicis brevis/m.opponens pollicis
" m.flexor carpi ulnaris
" m.interosseus dorsalis I/m.abductor digiti minimi

**Procedure**
*EMG*
" m.abductor pollicis brevis/m.opponens pollicis (truncus inferior)
" m.flexor carpi ulnaris (truncus inferior)
" m.interosseus dorsalis I/m.abductor digiti minimi
*Neurography*
" SCS: n.cutaneus antebrachii medialis bilaterally, n.cutaneus antebrachii lateralis

**References**
" Berry PR, Wallis WE. Venipuncture nerve injuries. Lancet 1977;1:1236-1237

**Revised**
" 1.7.1997 BF, 12.5.1997 BF

## MUSCULOCUTANEUS NERVE LESION

**Etiology**
" isolated lesions very rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets
" shoulder luxation

**Clinical features**
" weakness of elbow flexion
" loss biceps tendon reflex
" loss of sensation over the anterior lateral part of the forearm (n.cutaneus antebrachii lateralis)

**Strategy**
" demonstrate neurogenic EMG findings in the muscles innervated by n.musculocutaneus
" exclude other C5 or C6 radiculopathy, plexus brachialis lesion: lateral fascicle and plexus brachialis: upper trunk

**Expected abnormal findings**
*EMG*
" m.biceps brachii
" m.brachialis/m.coracobrachialis
*Neurography*
" SCS: n.cutaneus antebrachii lateralis

**Expected normal findings**
*EMG*

" m.deltoideus
" m.brachioradialis
" n.triceps

**Procedure**

*EMG*

" m.biceps brachii (n.musculocutaneus)
" m.brachioradialis (C6, plexus brachialis: upper trunk)
" m.flexor carpi radialis (plexus brachialis: lateral fascicle)
" m.deltoideus (C5/plexus brachialis: upper trunk)/m.supraspinatus/m.infraspinatus (plexus brachialis,: superior trunk)
" m.triceps (posterior fascicle)
" m.brachialis/m.coracobrachialis (n.musculocutaneus) optional

*Neurography*

" n.cutaneus antebrachii lateralis bilaterally

**References**

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**Revised**

" 15.4.1997

## AXILLARY NERVE LESION

**Etiology**

" luxation of the humerus
" fracture of the surgical neck of humerus
" traumatic injuries due to stabbing or bullets
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" an entrapment neuropathy “quadrilateral space syndrome “ has been described, we are doubtful about the existence of an entrapment

**Clinical features**

" weakness of shoulder abduction
" loss of sensation over the upper lateral part of the upper arm

**Strategy**

" demonstrate neurogenic EMG findings in the muscles innervated by the axillary nerve
" exclude other nerve lesions in the axilla, especially the suprascapular nerve

**Expected abnormal findings**

*EMG*

" m.deltoideus/m.teres minor

*Neurography*

" n.axillaris

**Expected normal findings**

*EMG*

" m.supraspinatus
" m.biceps
" m.triceps

**Procedure**

*EMG*

" m.deltoideus (n.axillaris)
" m.biceps brachii
" m.triceps
" m.supraspinatus/m.infraspinatus
" m.interosseus dorsalis I
" m.abductor pollicis brevis

*Neurography*

" n.axillaris MCS bilaterally

**References**

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**Revised**

" 15.4.1997

## SUPRASCAPULAR NERVE LESION

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" entrapment at the incisura scapulae
" entrapment at the spinoglenoid notch
" fractures of the scapula
" traumatic injuries to the shoulder

**Clinical features**

" weakness of upper arm abduction
" weakness of upper arm external rotation
" atrophy of m.supraspinatus and m.infraspinatus

" no sensory abnormalities

**Strategy**

" show neurogenic EMG findings in muscles innervated by n.suprascapularis
" localize lesion to incisura scapulae or the spinoglenoid notch

**Expected abnormal findings**

*EMG*

" neurogenic EMG findings in m.infraspinatus and m.supraspinatus if lesion is at incisura scapulae
" neurogenic EMG findings in only m.infraspinatus if lesion is at spinoglenoid notch

*Neurography*

" MCS n.suprascapularis: prolonged latency from Erb's point to m.infraspinatus/supraspinatus depending on the site of the lesion

**Expected normal findings**

*EMG*

" m.trapezius
" m.deltoideus
" m.rhomboideus
" m.biceps brachii

**Procedure**

*EMG*

" m.trapezius
" m.serratus anterior
" m.deltoideus
" m.infraspinatus
" m.supraspinatus
" m.rhomboideus

*Neurography*

" MCS n.suprascapularis: latency from Erb's point to m.infraspinatus and m.supraspinatus (use needle electrodes for recording and make sure you don't stimulate n.accessorius; bilateral study may be helpful)

**References**


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**Revised**

" BF 15.4.1997

## LONG THORACIC NERVE LESION

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" resection of first rib
" traumatic injuries, especially due to stabbing or bullets, sometimes blunt injuries

**Clinical features**

" winging of the scapula

**Strategy**

" show neurogenic EMG findings in m.serratus anterior

**Expected abnormal findings**

*EMG*

" neurogenic EMG findings in m.serratus anterior

**Expected normal findings**

*EMG*

" m.trapezius
" m.deltoideus

**Procedure**

*EMG*

" m.trapezius
" m.serratus anterior
" m.deltoideus
" m.infraspinatus
" m.rhomboideus
" m.biceps brachii

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**Revised**

" 15.4.1997

## DORSAL SCAPULAR NERVE LESION

**Etiology**
" isolated lesions very rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets

**Clinical features**
" slight winging of the scapula
" the medial margin of the scapula is displaced laterally
" the inferior angle of the scapula is rotated outwards

**Strategy**
" show neurogenic EMG findings in m.levator scapulae and m.rhomboideus major and minor
" exclude C5 radiculopathy and plexus brachialis lesion: upper trunk

**Expected abnormal findings**
*EMG*
" m.rhomboideus
" m.levator scapulae

**Expected normal findings**
*EMG*
" m.supraspinatus
" m.deltoideus

**Procedure**
*EMG*
" m.levator scapulae
" m.rhomboideus
" m.deltoideus
" m.supraspinatus
" m.biceps brachii
" m.triceps brachii

**Revised**
" 23.4.1997 BF

## THORACODORSAL NERVE LESION

**Etiology**
" isolated lesions very rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets

**Clinical features**
" weakness of shoulder adduction and inward rotation

**Strategy**
" show neurogenic EMG findings in m.latissimus dorsi
" exclude C7 radiculopathy and plexus brachialis lesion: middle trunk

**Expected abnormal findings**
*EMG*
" neurogenic EMG findings in m.latissimus dorsi

**Expected normal findings**
*EMG*
" m.latissimus dorsi

**Procedure**
*EMG*
" m.latissimus dorsi
" m.deltoideus
" m.triceps
" m.biceps brachii
" m.supraspinatus

**Revised**
" 23.4.1997 BF

## SUBSCAPULAR NERVE LESION

**Etiology**
" isolated lesions very rare
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" traumatic injuries due to stabbing or bullets

**Clinical features**
" weakness of inward rotation o0f the upper arm

**Strategy**
" show neurogenic EMG findings in m .subscapularis and m.teres major
" exclude C5 radiculopathy and plexus brachialis lesion: upper trunk

**Expected abnormal findings**
*EMG*
" m.subscapularis
" m.teres major

**Expected normal findings**
*EMG*

" m.supraspinatus
" m.deltoideus
" m.serratus anterior

**Procedure**

*EMG*

" m.levator scapulae
" m.rhomboideus
" m.deltoideus
" m.supraspinatus
" m.biceps brachii
" m.serratus anterior
" m.teres major

**Revised**

" 23.4.1997 BF

## BRACHIAL PLEXUS LESIONS

**Etiology**

*Trauma*

" traffic accidents, these injuries are normally combined with lesions of other proximal structures (other trunks, root avulsions)
" in contact sports blow to the head or shoulder may cause a “burner” or “stinger”, which may be due to a plexus lesions or radiculopathy
" humerus luxations
" stab and bullet wounds
" birth injuries especially in large babies there may be a disproportion between the shoulders of the baby and the pelvis of the mother. Erb’s palsy affects the upper part of the plexus and Klumpke’s palsy affects the lower part of the plexus

*Temporary compression*

" during coronary by-pass surgery 5-7 % of patients get a plexus lesion, usually the lower trunk is affected
" during general anesthesia if the arm is supinated and abducted more than 90 degress
" acute compression due to backpacks, especially in soldiers

*Inflammatory or unknown causes*

" acute idiopathic brachial plexus neuropathy (neuralgic amyotrophy, ”neuritis”). There is often acute onset with pain lasting a few days to a few weeks. Any part of the plexus, including spinal roots may be affected. This may be triggered by infections, immunization, surgery, pregnancy, childbirth or trauma. Diabetes predisposes to this. There is a rare autosomal dominant hereditary *disorder, “hereditary acute brachial plexus neuropathy”* where this occurs.

*Related with cancer or cancer treatment*

" tumors (especially lung cancer and metastasis of the lymph nodes in the axilla)
" radiotherapy for cancer

*Chronic compression (often misleadingly called thoracic outlet syndrome, TOS)*

" anomalous cervical rib
" following fracture of the clavicle due to deformity compression
" there are several poorly defined syndromes that have been suggested: (1) pectoralis minor syndrome and (2) scalenus anticus syndrome; the evidence is not convincing

*Iatrogenic*

" complication of plexus anesthesia
" hematoma from transaxillary percutaneous angiograms
" complications of surgery for thoracic outlet syndrome

**Clinical features**

" weakness and sensory abnormalities depend on the part of the plexus that is affects
" depending on the etiology pain may be present

*Upper trunk*

" weakness and/or atrophy of shoulder abduction, elbow flexion, upper arm outward rotation
" loss of sensation over the lateral side of the upper arm and the thumb

*Middle trunk*

" weakness of elbow extension, wrist flexion and extension
" numbness and/or loss of sensation in fingers 2-3

*Lower trunk*

" weakness of elbow extension, wrist flexion and extension
" numbness and/or loss of sensation in fingers 2-3

*Posterior cord*

" weakness and atrophy of muscles innervated by n.axillaris and n.radialis
" loss of sensation on the dorsal side of the hand between digits I and II

*Lateral cord*

" weakness and atrophy of muscles innervated by n.medianus and n.musculocutaneus
" numbness and/or loss of sensation of fingers 1-3

*Medial cord*

" weakness of muscles innervated by n.ulnaris and n.medianus
" numbness and/or loss of sensation in fingers 4-5

**Strategy**

" demonstrate neurogenic EMG findings in affected part of the plexus
" demonstrate abnormal SCS responses in affected parts
" exclude radiculopathy

**Expected abnormal and normal findings**

| Muscle | superior trunk | medial trunk | inferior trunk | lateral fascicle | posterior fascicle | medial fascicle | C5 | C6 | C7 | C8 | T1 |
|---|---|---|---|---|---|---|---|---|---|---|---|

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| m.rhomboideus | + | | | | | | ++ | | | | |
| m.supraspinatus | + | | | | | | ++ | | | | |
| m.infraspinatus | + | | | | | | ++ | | | | |
| m.deltoideus | + | | | | + | | ++ | + | | | |
| m.biceps brachii | + | | | + | | | + | ++ | | | |
| m.brachioradialis | + | | | | + | | | ++ | | | |
| m.triceps brachii | | + | | | + | | | | ++ | | |
| m.pronator teres | + | | | + | | | | + | ++ | | |
| m.flexor carpii radialis | | + | | + | | | | | ++ | | |
| m.latissimus dorsi | | + | | | + | | | | ++ | | |
| m.extensor digitorum | | + | | | + | | | | ++ | + | |
| m.extensor indicis proprius | | | + | | + | | | | | ++ | |
| m.interosseus dorsalis I | | | + | | | + | | | | ++ | + |
| m.abductor pollicis brevis | | | + | | | + | | | | + | ++ |
| paravertebral muscles | | | | | | | + | + | + | + | + |
| **SCS** | | | | | | | | | | | |
| n.medianus to digit I | + | | | | | | | | | | |
| n.medianus to digit II-III | | + | | | | | | | | | |
| n ulnaris to digit V | | | + | | | + | | | | | |
| n.radialis to digit I | | + | | | + | | | | | | |
| n.cutaneus antebrachii lateralis | + | | | + | | | | | | | |
| n cutaneus antebrachii medialis | | | + | | | | | | | | |
| H-reflex to m.flexor carpii rad. | | + | | + | | | | | + | | |

**Procedure**

*EMG*

" paravertebral muscles C5-Th1
" m.rhomboideus
" m.infraspinatus/m.supraspinatus
" m.deltoideus
" m.biceps brachii
" m.triceps
" m.pronator teres/m.flexor carpii radialis
" m.extensor indicis proprius
" m.interosseus dorsalis
" m.opponens pollicis/m.abductor pollicis brevis

*Neurography*

*SCS*

" n.medianus to digit I
" n.medianus to digits II and III
" n.ulnaris to digit V
" n.radialis

" n.cutaneus antebrachii lateralis
" n.cutaneus antebrachii medialis

*MCS*

" n.medianus
" n.ulnaris, fractionated (including plexus stimulation)
" n.medianus: H-reflex to m.flexor carpi radialis

**Note**

" Traumatic lesions of superior trunk may be combined with avulsion of cervical roots C7 - T1 (sometimes also C6), giving normal SNAP amplitudes in median, ulnar, and radial nerves (with loss of sensory function and cervical/cortical SEP) combined with loss of motor responses in the same nerves (lesions proximal to the sensory ganglion)

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**Revised**

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## PHRENIC NERVE LESIONS

**Etiology**

" acute idiopathic mononeuropathy
" surgery in the neck, especially sympathectomy
" as a complication of plexus anesthesia
" traumata

" malignant tumors
" intercostal drainage
" plexus anesthesia

**Clinical features**

" dyspnea
" weakness of respiratory muscles

**Strategy**

" demonstrate neurogenic EMG findings in m.diaphragma
" rule out C3 or C4 radiculopathy and affection of plexus cervicalis and plexus brachialis: upper trunk

**Expected abnormal findings**

*EMG*

" neurogenic EMG findings in m.diaphragma

*Neurography*

" MCS: n.phrenicus: reduced amplitude and prolonged latency

**Expected normal findings**

*EMG*

" m.trapezius (n.accessorius and plexus cervicalis)
" m.deltoideus/m.infraspinatus/m.supraspinatus (C5 and superior trunk)
" paravertebral muscles C3 and C4 (radiculopathy(

**Procedure**

*EMG*

" m.diaphragma
" m.trapezius (n.accessorius and plexus cervicalis)
" m.deltoideus/m.infraspinatus/m.supraspinatus (C5 and superior trunk)

*Neurography*

" MCS: n.phrenicus: reduced amplitude and prolonged latency

**References**

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## 5.2 CRANIAL NERVES

### OCULOMOTOR NERVE LESION (III)

**Etiology**

" acute idiopathic mononeuropathy
" head trauma
" congenital defects (Möbius syndrome)
" trauma during delivery to upper brachila plexus and cervical plexus
" neoplasms involving the brainstem nuclei, intracranial portion of the nerve (acoustic neuromas, meningeomas)
" vascular (especially in diabetes)
" aneurysm
" infections (meningitis, borreliosis, tuberculosis, herpes zoster, AIDS)
" sarcoidosis

**Clinical features**

" diplopia
" paralysis of external ocular muscles

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the oculomotor nerve
" exclude possibility of other cranial nerve lesions
" exclude ocular myopathy

**Expected abnormal findings**

*EMG*

" m.rectus superior
" m.rectus inferior
" m.rectus medialis
" m.obliqus inferior
" m.levator palpebrae superioris

**Expected normal findings**

*EMG*

" m.obliqus superior
" m.rectus lateralis

**Procedure**

*EMG*

" m.rectus lateralis (abducens nerve)
" m.obliqus superior (trochlear nerve)
" m.rectus superior/m.rectus inferior/m.rectus medialis/m.obliqus inferior (oculomotor nerve)
" m.levator palpebrae superior (oculomotor nerve)

**References**

" Rush JA, Younge BR: Paralysis of cranial nerves III, IV, and IV: Cause and prognosis in 1000 cases. Arch Ophthalmol 1981;99:76-

**Revised**

" 23.4.1997 BF

## TROCHLEAR NERVE LESION (IV)

**Etiology**

" acute idiopathic mononeuropathy
" head trauma
" neoplasms involving the brainstem nuclei, intracranial portion of the nerve (acoustic neuromas, meningeomas)
" vascular (especially in diabetes)
" aneurysm
" infections (meningitis, borreliosis, tuberculosis, herpes zoster, AIDS)
" sarcoidosis

**Clinical features**

" vertical diplopia combined with image tilting

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the oculomotor nerve
" exclude possibility of other cranial nerve lesions
" exclude ocular myopathy

**Expected abnormal findings**

*EMG*

" m.obliqus superior

**Expected normal findings**

*EMG*

" m.rectus lateralis
" m.rectus superior
" m.rectus inferior
" m.rectus medialis
" m.obliqus inferior
" m.levator palpebrae superioris

**Procedure**

*EMG*

" m.rectus lateralis (abducens nerve)
" m.obliqus superior (trochlear nerve)
" m.rectus superior/m.rectus inferior/m.rectus medialis/m.obliqus inferior (oculomotor nerve)
" m.levator palpebrae superior (oculomotor nerve)

**References**

" Rush JA, Younge BR: Paralysis of cranial nerves III, IV, and IV: Cause and prognosis in 1000 cases. Arch Ophthalmol 1981;99:76-

**Modified**

" 23.4.1997 BF

## TRIGEMINAL NERVE LESIONS (V)

**Etiology**

" Sjögren's syndrome, scleroderma, mixed connective tissue disease, systemic lupus erythematosus, rheumatoid arthritis
" Wegener's granulomatosis
" tumors: Intracranial or extracranial, metastatic or primary, meningioma, schwannoma, epidermoid, chordoma
" trauma (250)
" diabetes mellitus
" sinusitis
" herpes zoster (255)
" amyloidosis
" trauma
" surgery
" acute idiopathic mononeuropathy (idiopathic trigeminal neuropathy)
" neurinomas
" congenital trigeminal defects
" vascular (especially in diabetes)
" aneurysm
" infections (meningitis, borreliosis, tuberculosis, herpes zoster, AIDS)
" sarcoidosis
" facelifting surger may damage individual branches of n.trigeminus

**Clinical features**

" numbness and/or loss of sensation over the face
" weakness of closing of the jaw

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the trigeminal nerve
" demonstrate affection of the sensory branches of the trigeminal nerve
" exclude affection of other cranial nerves

**Expected abnormal findings**

*EMG*

" m.masseter/m.temporalis/m.tensor veli palatini

*Neurography*

" SCS: n.alveolaris inferior

*Blink reflex*
" n.supraorbitalis
" n.infraorbitalis
" n.mentalis
*Masseter reflex*

**Expected normal findings**

*EMG*
" m.orbicularis oris

**Procedure**

*EMG*
" m.masseter/m.temporalis/m.tensor veli palatini
" m.orbicularis oris
*Neurography*
" SCS: n.alveolaris inferior (optional)
*Blink reflex*
" n.supraorbitalis
" n.infraorbitalis
" n.mentalis
*Masseter reflex*

**References**

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**Revised**

" 23.4.1997 BF

## INFERIOR ALVEOLAR NERVE LESION (V)

**Etiology**

" extraction of tooth from lower jaw (usually wisdom's tooth)
" sagittal osteotomy
" during intraosseal tooth implantations
" tumors of the mandible

**Clinical features**

" numbness and/or loss of sensation of the skin below the lower lip

**Strategy**

" demonstrated conduction abnormality in the inferior alveolar nerve

**Expected abnormal findings**

*Neurography*
" inferior alveolar nerve SCS
*Blink reflex*
" abnormal finding with stimulation of the mental nerve

**Expected normal findings**

*EMG*
" m.masseter/m.temporalis
*Blink reflex*
" normal finding with stimulation of the infraorbital and supraorbital branches

**Procedure**

*EMG*
" m.masseter
*Neurography*
" n.alveolaris inferior bilaterally with near nerve needle electrodes

**References**

" Burt RK, Sharfman WH, Karp BI, Wilson WH: Mental neuropathy (numb chin syndrome). Cancer 19_70:877,

**Revised**

"

## LINGUAL NERVE LESION (V)

**Etiology**
" extraction of 2. or 3. molar teeth
" sagittal osteotomy
" tumors of the mandible

**Clinical features**
" loss of sensation and paresthesia over the tongue, floor of the mouth and medial side of gingiva,
" loss of taste over the anterior two thirds of the tongue

**Strategy**
" demonstrated conduction abnormality in the inferior alveolar nerve

**Expected abnormal findings**
*Neurography*
" inferior alveolar nerve SCS
*Blink reflex*
" abnormal finding with stimulation of the mental nerve

**Expected normal findings**
*EMG*
" m.masseter/m.temporalis
*Blink reflex*
" normal finding with stimulation of the infraorbital and supraorbital branches

**Procedure**
*EMG*
" m.masseter
*Neurography*
" n.alveolaris inferior bilaterally with near nerve needle electrodes

**References**
" Burt RK, Sharfman WH, Karp BI, Wilson WH: Mental neuropathy (numb chin syndrome). Cancer 19_70:877,

**Revised**
"

## ABDUCENS NERVE LESION (VI)

**Etiology**
" acute idiopathic mononeuropathy (especially in diabetes)
" vascular lesions in the nerve
" head trauma
" neoplasms involving the brainstem nuclei, intracranial portion of the nerve (acoustic neuromas, meningeomas)
" vascular lesions in the brainstem
" aneurysm
" infections (meningitis, borreliosis, tuberculosis, herpes zoster, AIDS)
" sarcoidosis
" Möbius syndrome (congenital weakness in muscles innervated by n.abducens and n.facialis, probably due to congenital agenesis of the brainstem nuclei)

**Clinical features**
" diplopia
" weakness of abduction lateral gaze)of the eye

**Strategy**
" demonstrate neurogenic EMG findings in muscles innervated by the oculomotor nerve
" exclude possibility of other cranial nerve lesions
" exclude ocular myopathy

**Expected abnormal findings**
*EMG*
" m.rectus lateralis

**Expected normal findings**
*EMG*
" m.obliqus superior
" m.rectus superior
" m.rectus inferior
" m.rectus medialis
" m.obliqus inferior
" m.levator palpebrae superior

**Procedure**
*EMG*
" m.rectus lateralis (abducens nerve)
" m.obliqus superior (trochlear nerve)
" m.rectus superior/m.rectus inferior/m.rectus medialis/m.obliqus inferior (oculomotor nerve)
" m.levator palpebrae superior (oculomotor nerve)

**References**
" Rush JA, Younge BR: Paralysis of cranial nerves III, IV, and IV: Cause and prognosis in 1000 cases. Arch Ophthalmol 1981;99:76-

**Modified**
" 23.4.1997 BF

## Etiology
- Bell's palsy
- polyradiculitis
- trauma
- herpes zoster, (Ramsay Hunt syndrome)
- borreliosis
- Möbius syndrome (congenital weakness in muscles innervated by n.abducens and n.facialis, probably due to congenital agenesis of the brainstem nuclei)
- tumors of the parotid gland
- surgery in the middle ear (cholesteatoma, stapedectomy, tympanoplasty etc), acustic neuroma, glomus jugulare tumours, parotid gland tumours, facielifting operations

## Clinical features
- weakness of the facial muscles
- inability close eyelids
- depending on the level of the lesion there may be loss of taste over the tongue
- numbness and loss of sensation of n.intermedius behind and around the ear

## Strategy
- demonstrate neurogenic EMG findings in muscles innervated by the facial nerve
- asses patophysiology (neurapraxia, axonal degeneration) and severity

## Expected abnormal findings
*EMG*
- m.orbicularis oculi/m.frontalis
- m.oribicularis oris/m.zygomaticus major

*Neurography*
- facial nerve MCS: reduced amplitude

*Blink reflex*
- delayed efferent path of the blink reflex

## Expected normal findings
*EMG*
- m.masseter

## Procedure
*EMG*
- m.orbicularis oculi/m.frontalis
- m.oribicularis oris/m.zygomaticus major

*Neurography*
- facial nerve MCS:
- r.temporalis: m.frontalis
- r.zygomaticus: m.nasalis
- r.buccalis: m.orbicularis oris
- r.mandibularis: m.depressor anguli oris
- r.cervicalis: m.platysma

*Blink reflex*

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**Revised**
" 23.4.1997, 12.5.1997 BF

## GLOSSOPHARYNGEAL NERVE LESION (IX)

**Etiology**
" processes around the jugular foramen (the vagus nerve and spinal accessory nerves are also affected)
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" isolated glossopharyngeal nerve lesions are very uncommon

**Clinical features**
" difficulties in swallowing
" numbness and loss of sensation of the pharynx

**Strategy**
" demonstrate neurogenic EMG findings in muscles innervated by the glossopharyngeal nerve

**Expected abnormal findings**
*EMG*
" m.stylopharyngeus

**Expected normal findings**
*EMG*
" m.genioglossus/m.hyoglossus
" m.trapezius
" m.chricothyreoideus

**Procedure**
*EMG*
" m.stylopharyngeus
" m.cricothyroideus (n.laryngeus superior)
" m.trapezius/m.sternocleidomastoideus (n.accessorius)
" m.genioglossus/m.hyoglossus (m.hypoglossus)

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**Revised**
" 23.4.1997

## VAGUS NERVE LESION (X)

**Etiology**
" surgery around the thyroid gland
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" vascular lesions
" processes around the jugular foramen n. glossopharyngeus and n. accessorius are also affected)

**Clinical features**
" numbness and loss of sensation of the external ear
" dysphagia
" dysartria

**Strategy**
" demonstrate neurogenic EMG findings in muscles innervated by the vagus nerve

**Expected abnormal findings**
*EMG*
" m.vocalis/m.crioarytenoideus
" m.cricothyroideus

" m.palatoglossus/m.palatopharyngeus

**Expected normal findings**

*EMG*

" m.trapezius/m.sternocleidomastoideus
" m.genioglossus/m.hyoglossus

**Procedure**

*EMG*

" m.vocalis/m.crioarytenoideus (intrinsic muscles of the larynx)
" m.cricothyroideus (n.laryngeus superior)
" m.palatoglossus/m.palatopharyngeus (n.vagus)
" m.trapezius/m.sternocleidomastoideus (n.accessorius)
" m.genioglossus/m.hyoglossus (m.hypoglossus)

**References**


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**Revised**

" 23.4.1997

## RECURRENT NERVE LESION (X)

**Etiology**

" surgery around the thyroid gland, carotid endarteractomy
" acute idiopathic mononeuropathy
" in association with systemic diseases: LED, AIDS

**Clinical features**

" dysartria

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the inferior laryngeal nerve

**Expected abnormal findings**

*EMG*

" m.vocalis/m.crioarytenoideus (intrinsic muscles of the larynx)

**Expected normal findings**

*EMG*

" m.cricothyroideus (innervated by n.laryngeus superior)

**Procedure**

" m.cricothyreideus
" m.vocalis/m.crioarytenoideus
" m.palatoglossus/m.palatopharyngeus

if clinically indicated

" m.sternocleidomastoideus
" m.genioglossus/m.hyoglossus (glossopharyngeal nerve)

**References**


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**Revised**

" 22.7.1997

## ACCESSORY NERVE LESION (XI)

**Etiology**

" lymph node biopsy, or excision
" surgical trauma, radical neck dissection, l
" blunt or penetrating trauma
" jugular vein cannulation
" cervical internal carotid artery dissection
" carotid endarterectomy
" radiation therapy to head and neck

" coronary artery bypass surgery
" neurinoma
" trauma to the neck
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" part of the foramen jugulare syndrome (glomus tumor) IX, X, XI.
" fracture of the scull base
" tumors of the scull base
" aneurysms of the carotid artery

**Clinical features**

" drooping of the shoulder
" winging of the scapula
" weakness of shoulder elevation and head rotation to the contralateral side

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the accessory nerve
" localize lesion (trapezius with or without sternocleidomastoideus lesion
" exclude other reason for atrophy

**Expected abnormal findings**

*EMG*

" m. trapezius
" m.sternocleidomastoideus is involved in proximal lesions (not denervated in accessory nerve lesions due to lymph node biopsies)

*Neurography*

" MCS: n.accessorius reduced amplitude

**Expected normal findings**

*EMG*

" m.masseter
" m.deltoideus
" m.orbicularis oris

**Procedure**

*EMG*

" m. trapezius (upper, middle and lower portion should be examined separately)
" m.sternocleidomastoideus
" m.hyoglossus/m.genioglossus
" m.masseter
" m.oribicularis oris/m.zygomaticus major

*Neurography (optional))*

" MCS: n.accessorius

**References**


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" 23.4.1997

## HYPOGLOSSAL NERVE LESION (XII)

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" fracture of the scull base
" tumors of the scull base and neck
" aneurysms of the carotid artery
" carotid endarterectomy
" clivus chordoma
" glomus jugulare tumor
" metastases to base of skull
" carotid artery aneurysm at base of skull
" atlantoaxial subluxation

" complication of radiation therapy
" neuroma

**Clinical features**

" isolated hypoglossal nerve lesions are very rare
" weakness and atrophy of the tongue

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by n.hypoglossus(mainly muscles of the tongue)
" differentiate from bulbar paralysis involving other cranial nerves

**Expected abnormal findings**

*EMG*

" m.hyoglossus/m.genioglossus

*Neurography*

" MCS: m.hyoglossus reduced amplitude

**Expected normal findings**

*EMG*

" m.masseter
" m.orbicularis oris

**Procedure**

*EMG*

" m.hyoglossus/m.genioglossus
" m.masseter
" m.oribicularis oris/m.zygomaticus major

*Neurography (optional, measurement requires special electrode)*

" MCS: n.hypoglossus

**References**

" DeCock M: Nervus hypoglossus (n. XII). Acta Otorhinolaryngol Belg 1986;40:260-
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**Revised**

" 23.4.1997 BF

## 5.3 LOWER EXTREMETIES

### SCIATIC NERVE LESION

**Etiology**

" surgery for hip joint replacement
" gluteal injection
" gluteal contusion
" nerve infarction
" fractures of the femur
" hip fracture and/or dislocation
" metastatic lesions
" radiation therapy

**Clinical features**

" weakness of muscles innervated by n.ischiadicus
" usually n.peroneus is more affected than n.tibialis
" numbness and/or loss of sensation in the foot
" pain may be present

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by n.ischiadicus
" exclude radiculopathy or other proximal lesion
" define level of lesion if possible

**Expected abnormal findings**

*EMG:*

neurogenic findings in 1-5 depending on the level of the lesion (examine one from each set of muscles)

" 1. biceps femoris caput brevis
" 2. semitendinosus/semimebranosus
" 3. tibialis anterior/peroneus longus/extensor hallucis longus
" 4. gastrocnemius caput mediale or laterale.
" 5. intrinsic foot muscles)

*Neurography:*

" low M-amplitude in m.extensor digitorum brevis, abductor hallucis
" F-wave prolonged or absent in n.peroneus or n.tibialis
" n.suralis SNAP amplitude reduced
" n.peroneus superficialis SNAP amplitude reduced
" H-reflex absent or prolonged

**Expected normal findings**

*EMG*

" paravertebral muscles
" gluteus maximus (n.gluteus inferior)

" tensor fascia latae/gluteus medius (n.gluteus superior)

**References**

"

**Revised**

" bf 15.4.1997

## LUMBOSACRAL PLEXUS LESION

**Etiology**

" often the lumbar and the sacral plexus are separately affected but some times both may be affected
" traumatic lesions in the hip region
" surgery for hip joint replacement
" injection into erroneous site
" tumors and metastatic lesions
" radiation therapy
" acute idiopathic mononeuropathy (neuralgic amyotrophy) (in diabetes patients often called diabetic amyotrophy)
" pelvic fractures
" aortic aneurysm
" pregnancy
" retroperitoneal hematoma

**Clinical features**

" weakness of muscles innervated by n.ischiadicus
" weakness of muscles innervated by n.femoralis and n.obturatorius
" usually n.peroneus is more affected than n.tibialis
" numbness and/or loss of sensation in the thigh, leg and foot
" pain may be present

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the lumbosacral plexus
" exclude radiculopathy

**Expected abnormal findings**

*EMG:*

neurogenic findings in muscles innervated by the lumbosacral plexus

" gluteus maximus (n.gluteus inferior)
" tensor fascia latae/gluteus medius(n.gluteus superior)
" biceps femoris caput brevis
" semitendinosus/semimebranosus
" tibialis anterior/peroneus longus/extensor hallucis longus
" gastrocnemius caput mediale or laterale.
" quadriceps femoris
" adductor magnus
" intrinsic foot muscles

*Neurography:*

" low M-amplitude in m.extensor digitorum brevis, m.abductor hallucis
" F-wave prolonged or absent in n.peroneus or n.tibialis
" n.suralis SNAP amplitude reduced
" n.saphenus SNAP amplitude reduced
" H-reflex absent or prolonged

**Expected normal findings**

*EMG*

" paravertebral muscles

**Procedure**

*EMG:*

examine one from each set of muscles

" m.gluteus maximus (n.gluteus inferior)
" m.tensor fascia latae/m.gluteus medius (n.gluteus superior)
" m.biceps femoris caput brevis
" m.semitendinosus/m.semimebranosus
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus
" m.gastrocnemius caput mediale or laterale.
" m.quadriceps femoris/m.iliopsoas
" m.adductor magnus

*Neurography*

" MCS: n.peroneus, n.tibialis
" SCS: n.suralis, n.peroneus superficialis, n.saphenus
" H-reflex

**References**


" Sander JE, and Sharp FR. Lumbosacral plexus neuritis. Neurology 1981:31;470-473
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**Revised**

" 8.5.1997 BF

## LUMBAR PLEXUS LESION

### Etiology
" acute idiopathic mononeuropathy (neuralgic amyotrophy) (in diabetes patients often called diabetic amyotrophy)
" traumata
" pelvic fractures
" aortic aneurysm
" pregnancy
" retroperitoneal hematoma
" tumors
" radiation

### Clinical features
" weakness of knee extension, thigh abduction and flexion
" numbness and/or loss of sensation over the anterior and medial side of the thigh
" loss of patellar tendon reflex

### Strategy
" demonstrate neurogenic EMG findings in muscles innervated by the lumbar plexus
" exclude radiculopathy

### Expected abnormal findings
*EMG*

neurogenic findings in
" m.quadriceps femoris(m.vastus lateralis/m.vastus medialis or m.recuts femoris)
" tibialis anterior
" iliopsoas
" adductor magnus

*Neurography*
" n.saphenus SNAP amplitude reduced

### Expected normal findings
*EMG*
" paravertebral muscles
" m.gluteus maximus (n.gluteus inferior)
" m.tensor fascia latae/gluteus medius(n.gluteus superior)
" m.biceps femoris caput brevis
" m.semitendinosus/m.semimebranosus
" m.gastrocnemius caput mediale or laterale
" intrinsic foot muscles

*Neurography*
" n.tibialis and n.peroneus to m.extensor digitorum brevis normal
" n.suralis normal
" H-reflex normal

### Procedure
*EMG:*

examine one from each set of muscles
" m.tensor fascia latae/gluteus medius(n.gluteus superior)
" m.tibialis anterior/peroneus longus/extensor hallucis longus
" m.gastrocnemius caput mediale or laterale.
" m.quadriceps femoris
" m.adductor magnus
" m.iliopsoas

*Neurography*
" MCS: n.peroneus, n.tibialis
" SCS: n.suralis, n.peroneus superficialis, n.saphenus
" H-reflex

### References
" Sander JE, and Sharp FR. Lumbosacral plexus neuritis. Neurology 1981:31;470-473
" Awerbuch GI, Nigro MA, Sandyk R, Levin JR.: Relapsing lumbosacral plexus neuropathy. Eur Neurol 1991;31:348-351Bradley WG, Chad D, Verghese JP, Liu HC, Good P, Gabbai AA, Akelman LS. Painful lumbosacral plexopathy with elevated erythrocyte sedimentation rate: a treatable inflammatory syndrome. Ann Neurol 1984;5:457-464
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" Evans BA, Stevens JC, Dyck PJ. Lumbosacral plexus neuropathy. Neurology 1981;31:1327-1330.

### Revised
" 8.5.1997 BF

## SACRAL PLEXUS LESION

### Etiology
" traumatic lesions in the hip region
" surgery for hip joint replacement
" injection into erroneous site
" tumors and metastatic lesions
" radiation therapy

" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" aortic aneurysm
" pregnancy

**Clinical features**

" weakness of muscles innervated by n.ischiadicus
" usually n.peroneus is more affected than n.tibialis
" numbness and/or loss of sensation in the foot
" pain may be present

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the sacral plexus
" exclude radiculopathy

**Expected abnormal findings**

*EMG*

neurogenic findings in muscles innervated by the lumbosacral plexus

" m.gluteus maximus (n.gluteus inferior)
" m.tensor fascia latae/m.gluteus medius(n.gluteus superior)
" m.biceps femoris caput brevis
" m.semitendinosus/m.semimebranosus
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus
" m.gastrocnemius caput mediale or laterale.
" m.quadriceps femoris
" (m.adductor magnus) this muscle may allso be innervated from n.ischiaducus
" intrinsic foot muscles

*Neurography*

" low M-amplitude in EDB, abductor hallucis
" F-wave prolonged or absent in n.peroneus or n.tibialis
" n.suralis SNAP amplitude reduced
" H-reflex absent or prolonged

**Expected normal findings**

*EMG*

" paravertebral muscles

*Neurography*

" SCS: n.saphenus

**Procedure**

*EMG:*

examine one from each set of muscles

" m.gluteus maximus (n.gluteus inferior)
" m.tensor fascia latae/m.gluteus medius(n.gluteus superior)
" m.biceps femoris caput brevis
" m.semitendinosus/m.semimebranosus
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus
" m.gastrocnemius caput mediale or laterale.
" m.quadriceps femoris
" m.adductor magnus

*Neurography*

" MCS: n.peroneus, n.tibialis
" SCS: n.suralis, n.peroneus superficialis, n.saphenus
" H-reflex

**References**

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**Revised**

" 8.5.1997

## PUDENDAL NERVE LESION

**Etiology**

" pelvic tumors
" pelvic fractures and surgery for pelvic fractures
" surgery
" perineal branches may by compressed during cycling

**Clinical features**

" urinary bladder dysfunction, mainly weakness of the bladder contractility
" anal incontinence, weakness of the sphincter anii externus
" impotence
" numbness and/or loss of sensation around the perianal region and in the penis

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the pudendal nerve

**Expected abnormal findings**

*EMG*
" m.sphincter ani externus
" m.bulbocavernosus
" m.ishiocavernosus
*Neurography*
" n.pudendus
" n.dorsalis penis in men
*SEP*
" pudendal SEP

**Expected normal findings**
*EMG*
" m.gluteus maximus

**Procedure**
*EMG*
" m.sphincter ani externus
" m.puborectalis (innervated form the pudendal plexus)
" m.gastrocnemius (n.tibialis)
" m.tibialis anterior n.peroneus
*SFEMG*
" fibre denisity in m.sphincter anii externus
*Neurography*
" SCS: n.dorsalis penis in men
" MCS: n.pudendus using the special St Marks electrode
*SEP*
" pudendal SEP, stimulation should be unilateral
*Reflexes*
" bulbocavernosus reflex

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**Revised**
" 16.4.1997

## LESION OF THE LATERAL CUTANEOUS NERVE OF THE THIGH (MERALGIA PARESTHETICA)

**Etiology**
" entrapment at the anterior superior iliac spine
" surgery to the anterior iliac crest (bone transplantation)
" surgery around the anterior iliac spine

**Clinical features**
" numbness and/or loss of sensation over the anterior-lateral part of the thigh
" sometimes pain

**Strategy**
" demonstrate abnormality of the lateral cutaneous nerve of the thigh at the superior anterior iliac spine

**Expected abnormal findings**
*Neurography*
" lateral cutaneous nerve of the thigh SCS abnormal at the anterior superior iliac spine

**Expected normal findings**
*EMG*
" m.iliopsoas
" m.adductor magnus
" m.vastus lateralis

**Procedure**
*Neurography*
" SCS: n.cutaneus femoris lateralis, ortodromically with near nerve needle electrodes
*EMG*
" m.iliopsoas
" m.adductor magnus
" m.vastus lateralis

**References**
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**Revised**
" 6.4.1997, 12.4.1997

## ILIOINGUINAL NERVE LESION

**Etiology**
" retroperitoneal tumors
" surgery, especially herniorraphy and kidney operations
" acute idiopathic mononeuropathy ("neuralgic amyotrophy, neuritis")
" entrapment has been described as the nerve emerges thorough the abdominal wall

**Clinical features**
" weakness of the lower abdominal muscles, bulging of the abdomen
" numbness and/or loss of sensation over the symphysis, labia majora (in women), base of the penis and scrotum (in men)

**Strategy**
" demonstrate neurogenic EMG findings in m.obliqus internus abdominis
" differentiate from ilioinguinal nerve and genitofemoral nerve lesions

**Expected abnormal findings**
*EMG*
" m.obliqus internus abdominis
*SEP*
" SEP by stimulation of the sensory areas innervated by the nerve

**Expected normal findings**
*EMG*
" m.iliopsoas
" m.rectus abdominis

**Procedure**
*EMG*
" m.obliqus internus abdominis
" m.iliopsoas (femoral nerve, L2-L4 radiculopathy)
*Neurography*
" sometimes it is possible to record SCS with needle electrodes
*SEP*
" SEP by stimulation of the sensory areas innervated by the nerve

**References**
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**Revised**
" 6.4.1997 BF

## ILIOHYPOGASTRIC NERVE LESION

**Etiology**
" retroperitoneal tumors
" surgery for inguinal hernia, especially kidney
" acute idiopathic mononeuropathy ("neuralgic amyotrophy, neuritis")
" the anterior sensory branch can be damaged by Pfannenstiel's incision

**Clinical features**
" weakness of the lower abdominal muscles, bulging of the abdomen
" numbness and/or loss of sensation below and behind the spina ilica superior

**Strategy**
" demonstrate neurogenic EMG findings in m.obliqus internus abdominis
" differentiate from ilioinguinal nerve and genitofemoral nerve lesions

**Expected abnormal findings**
*EMG*
" m.obliqus internus abdominis
*SEP*
" SEP by stimulation of the sensory areas innervated by the nerve: lateral cutaneous branch and anterior cutaneous branch

**Expected normal findings**
*EMG*
" m.iliopsoas
" m.rectus abdominis
*Neurography*
" SCS: n.ilioinguinalis

**Procedure**
*EMG*
" m.obliqus internus abdominis
" m.iliopsoas (femoral nerve, L2-L4 radiculopathy)
*SEP*
" SEP by stimulation of the sensory areas innervated by the nerve: lateral cutaneous branch and anterior cutaneous branch

*Neurography*
" SCS: n.ilioinguinalis

**References**
" Stulz P, Pfeiffer KM. Peripheral nerve injuries resulting form common surgical procedures in the lower portion of the abdomen. Arch Surg 1982;117:324-327

**Revised**
" 6.4.1997

## GENITOFEMORAL NERVE LESION

**Etiology**
" herniorraphy and other operations in the lateral pelvis or inguinal ligament
" acute idiopathic mononeuropathy ("neuralgic amyotrophy, neuritis")

**Clinical features**
" numbness and/or loss of sensation medially below the inguinal ligament
" abnormal cremaster reflex

**Strategy**
" there are no simple reliable nerve conduction methods to study this nerve
" exclude ilioinguinal nerve lesion and iliohypoastric lesion

**Expected abnormal findings**
*Special studies*
" abnormal cremaster reflex
" n.genitofemoralis

**Expected normal findings**
*EMG*
" transverse abdominal muscles
*Neurography*
" SCV: n.ilioinguinalis
*SEP*
" n.ilioinguinalis
" n.iliohypogastricus

**Procedure**
*Special studies*
" abnormal cremaster reflex
*EMG*
" transverse abdominal muscles
*Neurography*
" SCV: n. ilioinguinalis
*SEP*
" n.genitofemoralis
" n.ilioinguinalis
" n.iliohypogastricus

**References**
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**Revised**
" 6.4.1997

## FEMORAL NERVE LESION

**Etiology**
" trauma in the inguinal region
" surgery: appendectomy, hip operations, herniorraphy, hysterectomy through Pfannenstiel's incision
" arteriography through the femoral artery
" femoral artery reconstruction
" hematoma in the groin, retroperitoneal hematoma
" metastasis in the retroperitoneal region
" retroperitoneal abscess
" aneurysm of the femoral artery
" acute idiopathic mononeuropathy (neuralgic amyotrophy)

**Clinical features**
" weakness of knee extension
" atrophy of m.quadriceps femoris
" loss patellar tendon reflex
" numbness and/or loss of sensation over the anterior part of the thigh and anterior-medial part of the leg

**Strategy**
" demonstrate neurogenic EMG findings in muscles innervated by the femoral nerve and abnormal SNC findings in n.saphenus and r.cutaneus femoris anterior
" differentiate from lumbar plexus lesion and L2-L4 radiculopathy

**Expected abnormal findings**
*EMG*
" m.quadriceps femoris
" m.iliopsoas if the lesion is above the inguinal ligament
*Neurography*
" abnormal SNC in n.saphenus

" abnormal SNC in r.cutaneus femoris anterior

**Expected normal findings**

*EMG*

" m.tibialis anterior
" m.adductor magnus

*Neurography*

" SCS: n.suralis

**Procedure**

*EMG*

" m.iliopsoas
" m.vastus lateralis/m.vastus medialis
" m.adductor magnus (obturator nerve, L2-L3 radiculopathy)
" m.tilbialis anterior (peroneal nerve, L4 radiculopathy)
" paravertebral muscles L2-L4 (radiculopathy)

*Neurography*

" SNC: n.saphenus
" SNC: r.cutaneus femoris anterior (optional)

**References**


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**Revised**

" 7.4.1997 BF

## SAPHENOUS NERVE LESION

**Etiology**

" iatrogenic lesion following surgery for varicose veins

**Clinical features**

" loss of sensation over the anterior-medial part of the thigh

**Strategy**

" demonstrate abnormality of n.saphenus
" exclude proximal nerve lesions, radiculopathy L3 or L4, and lumbar plexus lesions

**Expected abnormal findings**

*Neurography*

" saphenous nerve SCS

**Expected normal findings**

*EMG*

" m.tibialis anterior
" m.vastus lateralis/medialis
" m.adductor magnus

**References**


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**Revised**

" 6.4.1997 BF

## OBTURATOR NERVE LESION

**Etiology**

" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" pelvic fractures
" complication of hip replacement
" obturator hernia
" metastasis of the bones around the obturator foramen

**Clinical features**

" weakness of thigh adduction
" loss of sensation or numbness on the inner side of the thigh

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the obturator nerve
" differentiate from L2-L4 radiculopathy and lumbar plexus lesion

**Expected abnormal findings**

*EMG*

" m.adductor magnus/m.adductor longus/m.gracilis

*SEP*

" n.obturatorius

**Expected normal findings**

*EMG*

" m.vastus lateralis/m.vastus medialis
" m.iliopsoas
" m.tibialis anterior
" paravertebral muscles L2-L4

**Procedure**

*EMG*

" m.adductor magnus/m.adductor longus
" m.vastus lateralis/m.vastus medialis
" m.iliopsoas
" m.tibialis anterior
" m.tensor fascia latae
" paravertebral muscles L2-L4

*SEP*

" stimulation of the skin area innervated by the cutaneous branch of the obturator nerve

**Note**

" m.adductor magnus may be innervated also from the sciatic nerve
" m.adductor longus may be innervated from the femoral nerve

**References**


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**Revised**

" 16.4.1997 BF

## POSTERIOR CUTANEOUS FEMORAL NERVE LESION

**Etiology**

" lacerations over the posterior part of the thigh
" acute idiopathic mononeuropathy (neuralgic amyotrophy)
" injections into the buttock
" pelvic fractures
" metastatic tumors

**Clinical features**

" numbness and/or loss of sensation over the dorsal side of the thigh

**Strategy**

" demonstrate abnormal findings in n.cutaneus femoris posterior
" differentiate from sciatic nerve lesions

**Expected abnormal findings**

*Neurography*

" SCS: n.cutaneus femoris posterior

**Expected normal findings**

*EMG*

" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior
" paravertebral muscles L4-S1

*Neurography*

" SCS: n.suralis
" MCS: n.peroneus profundus, n.tibialis

**Procedure**

*EMG*

" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior
" m.gastrocnemius
" m.biceps femoris
" paravertebral muscles L4-S1

*Neurography*

" SCS : n.cutaneus femoris posterior, n.suralis
" MCS: n.tibialis, n.peroneus

**References**


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**Revised**

" 16.4.1997

## COMMON PERONEAL NERVE LESION AT THE KNEE

**Etiology**

" trauma to the lateral side of the knee
" prolonged kneeling, when picking berries
" inversion distorsion of the ankle
" prolonged pressure to the fibular head, e.g. when sitting with knees crossed

" surgery around the knee

**Clinical features**

" weakness of ankle dorsiflexion and extension of toes
" loss of sensation or numbness over the dorsal side of the foot and antero-lateral part of the leg

**Strategy**

" demonstrate neurogenic EMG findings in muscles innervated by the common peroneal nerve
" demonstrate conduction abnormality across fibular head
" exclude proximal lesion

**Expected abnormal findings**

*EMG*

" m.tibialis anterior
" m.extensor hallucis longus
" m.peroneus longus
" m.peroneus brevis

*Neurography*

" n.peroneus profundus MCS across fibular head abnormal: conduction block or conduction velocity reduced
" n.peroneus superficialis SNAP amplitude reduced

**Expected normal findings**

*EMG:*

" m.tibialis posterior
" m.flexor hallucis longus
" m.biceps femoris caput brevis
" m.semitendinosus
" m.gluteus medius
" m.tensor fascia latae
" paravertebral muscles L5

*Neurography*

" SCS: n.suralis
" MCS: n.tibialis

**Procedure**

*EMG*

" m.tibialis anterior/m.extensor hallucis longus (n.peroneus profundus)
" m.peroneus longus/m.peroneus brevis neurogenic (n.peroneus superficialis)
" m.tibialis posterior/m.flexor hallucis longus (n.tibialis, L5)
" m.gluteus medius/m.tensor fascia latae (L5)
" m.biceps femoris caput brevis (optional, sciatic nerve)

*Neurography*

" MCS: n.peroneus profundus across fibular head and below the knee (if no response or very small response is recorded from m.extensor digitorum brevis the measurement should be made with surface electrodes over m.tibialis anterior)
" SCS: n.peroneus superficialis

**Note**

" In most people m.extensor digitorum brevis is innervated solely by n.peroneus profundus. In a few persons the muscle receives innervation from a branch of n.peroneus superficialis (n.peroneus accessorius)

**References**


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**Revised**
" 3.4.1997

## ANTERIOR TARSAL TUNNEL SYNDROME

**Etiology**
" compression of the distal part of n.peroneus profundus at the ankle

**Clinical features**
" loss of sensation over the dorsal side of toes I and II
" atrophy of m.extensor digitorum brevis

**Strategy**
" demonstrate lesion of the distal part of n.peroneus profundus
" exclude proximal lesion of the peroneal nerve

**Expected abnormal findings**
*Neurography*
" abnormal SCS of n.peroneus profundus (toes to ankle)
*EMG*
" m.extensor digitorum brevis

**Expected normal findings**
*Neurography*
" SCS: n.suralis
" SCS: n.peroneus superficialis
*EMG*
" m.tibialis anterior/m.extensor hallucis longus

**Strategy**
*EMG*
" m.tibialis anterior/m.extensor hallucis longus
" m.extensor digitorum brevis
" m.peroneus longus/m.peroneus brevis
*Neurography*
" SCS: n.peroneus superficialis both branches
" SCS: n.suralis
" SCS: n.peroneus profundus
" MCS: n.peroneus

**References**
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**Revised**
" 6.4.1997 BF, 8.5.1997 BF

## SUPERFICIAL PERONEAL NERVE LESION IN THE FOOT

**Etiology**
" severe distorsion of the ankle
" surgery to the forefoot or lover part of the leg
" traumatic herniation of muscle trough fascia
" usually a lesion of the laterally located branch of the terminal sensory branches

**Clinical features**
" loss of sensation on the dorsal side of the foot pain in the forefoot

**Strategy**
" demonstrate lesion of the distal superficial peroneal nerve branch

" exclude proximal lesion of the peroneal nerve

**Expected abnormal findings**

*Neurography*

" abnormal SCS of the lateral branch of n.peroneus superficialis

**Expected normal findings**

*Neurography*

" SCS: n.suralis
" SCS: medial branch of n.peroneus superficialis

*EMG*

" m.tibialis anterior

**Strategy**

*EMG*

" m.tibialis anterior/m.extensor hallucis longus

*Neurography*

" SCS: n.peroneus superficialis both branches
" SCS: n.suralis
" MCS: n.peroneus

**References**

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**Revised**

" BF 8.5.1997

## TARSAL TUNNEL SYNDROME

**Etiology**

" entrapment neuropathy of the tibial nerve or its branches in the area of the medial malleolus under the flexor retinaculum
" usually due to rheumatoid arthritis, tenosynovitis or malleolar fractures

**Clinical features**

" very rare
" numbness and pain over the sole of the foot
" pain around the medial malleolus

**Strategy**

" demonstrate neurogenic EMG findings in intrinsic foot muscles innervated by the lateral and medial plantar nerve
" demonstrate reduced CV in the tibial nerve across the tarsal tunnel
" differentiate from proximal tibial nerve lesion

**Expected abnormal findings**

*EMG*

" m.abductor hallucis
" m.abductor digiti minimi

*Neurography*

" MCS: reduced CV or prolonged DL in n.tibialis across the tarsal tunnel
" SCS: reduced CV in n.tibialis across the tarsal tunnel

**Expected normal findings**

*EMG*

" m.gastrocnemius c.mediale/laterale
" m.tibialis anterior

*Neurography*

" SCS: n.suralis
" MCS: n.peroneus

**Procedure**

*EMG*

" m.abductor hallucis
" m.abductor digiti minimi
" m.extensor digitorum brevis
" m.gastrocnemius c.mediale/laterale
" m.tibialis anterior

*Neurography*

" MCS: n.tibialis across the tarsal tunnel (use near nerve needles for stimulation)
" SCS: n.tibialis across the tarsal tunnel (use near nerve needles for recording)

**Note**

" although this is often suspected this is a *very rare* disorder, we have never seen an idiopathic tarsal tunnel syndrome without deformity of the ankle around the medial malleolus
" the intrinsic foot muscles show mild to moderate neurogenic abnormalities even in healthy subjects, compare findings with the opposite foot

**References**

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" Felsenthal G, Butler DH, Shear MS. Axross tarsal tunnel motor nerve conduction technique. Arch Phys Med Rehabil 1992;73:64-69
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**Revised**
" 6.4.1997 BF

## MORTON´S METATARSALGIA

**Etiology**
" entrapment of the plantar digital nerves between the distal metatarsal heads
" usually the digital nerves II and III, between the II/III and III/IV metatarsal heads, are affected

**Clinical features**
" a fairly common disorder in 50-70 year old women, sometimes seen in younger persons
" pain in the forefoot when walking, often symptoms are alleviated if shoes are taken off
" clinically often distinct painful area between affected metatarsal heads
" associated with hallux valgus and rheumatoid arthritis
" plantar digital nerve to interspaces II/III and III/IV commonly affected, in most patients both interspaces affected

**Strategy**
" demonstrate lesion of the plantar digital nerves

**Expected abnormal findings**
*Neurography*
" SCS: educed CV of the affected plantar digital nerves
" SCS: reduced sensory AMPL of the affected plantar digital nerves

**Procedure**
" orthodromic SCS with near nerve needle electrodes in all four interspaces (I/II, II/III, III/IV and IV/V)
" either lateral or medial sides of toes can be studied
" in borderline cases it may be helpful to study both lateral and medial plantar digital branches

**Note**
" in most patients the digital nerves II (between II/III metatarsal heads) and III (between III/IV metatarsal heads) are affected

**References**
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**Revised**
" **6.4.1997**

## SURAL NERVE LESION AT THE KNEE

**Etiology**
" Baker's cyst
" surgery, arthroscopy of the knee
" knee trauma

**Clinical features**
" loss of sensation over the lateral side of the foot
" sometimes pain

**Strategy**
" demonstrate lesion of n.suralis
" exclude proximal lesion, especially S1 radiculopathy unless the lesion is obvious

**Expected abnormal findings**
*Neurography*
" SCS: n.suralis

**Expected normal findings**
*EMG*
" m.gastrocnemius
" n.tibialis innervated intrinsic muscles of the foot
*Neurography*
" n.peroneus superficialis

**Procedure**
*EMG*
" m.gastrocnemius c. mediale
" optional : m.abductor digiti minimi, adductor hallucis
*Neurography*
" SCS: n.suralis, n.peroneus superficialis
" MCS: n.tibialis

**References**
" Smith BE, Litchi WJ. Sural mononeuropathy: a clinical and electrophysiological study. Neurology 1989;39:296

**Modified**
" 6.4.1997 BF

## SURAL NERVE LESION IN THE CALF

**Etiology**
" compression by ski boot or tight socks

" surgery, varicose veins

**Clinical features**

" loss of sensation over the lateral side of the foot
" sometimes pain

**Strategy**

" demonstrate lesion of n.suralis
" exclude proximal lesion, especially S1 radiculopathy unless the lesion is obvious

**Expected abnormal findings**

*Neurography*

" SCS: n.suralis

**Expected normal findings**

*EMG*

" m.gastrocnemius
" n.tibialis innervated intrinsic muscles of the foot

*Neurography*

" n.peroneus superficialis

**Procedure**

*EMG*

" m.gastrocnemius c. mediale
" optional : m.abductor digiti minimi, adductor hallucis

*Neurography*

" SCS: n.suralis, n.peroneus superficialis
" MCS: n.tibialis

**References**

" Gross JA, Hamilton WJ, Swift TR. Isolated mechanical lesions of the sural nerve. Muscle Nerve 1980;3:248-249
" Shaffrey ME, Jane JA, Persing JA, Shaffrey CI, Phillps LH. Surgeon's foot: a report of sural nerve palsy. Neurosurgery 1992;30:927-930

**Modified**

" 6.4.1997 BF

### SURAL NERVE LESION AT THE MALLEOLUS

**Etiology**

" bimalleolar fracture
" compression by ski boot
" ankle sprain
" cysts
" posttraumatic fibrosis
" surgery around the ankle
" the sural nerve is often used for nerve biopsy and nerve grafting

**Clinical features**

" loss of sensation over the lateral side of the foot
" sometimes pain

**Strategy**

" demonstrate lesion of n.suralis
" exclude proximal lesion, especially S1 radiculopathy unless the lesion is obvious

**Expected abnormal findings**

*Neurography*

" SCS: n.suralis

**Expected normal findings**

*EMG*

" m.gastrocnemius
" n.tibialis innervated intrinsic muscles of the foot

*Neurography*

" n.peroneus superficialis

**Procedure**

*EMG*

" m.gastrocnemius c. mediale
" optional : m.abductor digiti minimi, adductor hallucis

*Neurography*

" SCS: n.suralis (bilaterally), n.peroneus superficialis
" MCS: n.tibialis

**References**

" Pringle RM. Protheroe K, Mukherjee CI. Entrapment neuropathu of the sural nerve. J Bone Join Surg [Br](Br) 1974;56B:465-468
" Perlman MD. Os peroneum fracutre with sural nerve entrapment neuritis. J Foot Surg 1990;29:119-121

**Modified**

" 6.4.1997 BF

## 6. RADICULOPATHIES

**General strategy**

" demonstrate neurogenic EMG findings in muscles innervated by specific myotomes
" the EMG finding is definite if muscles innervated by the ventral ramus and dorsal ramus are affected. Therefore neurogenic abnormalities should be demonstrated in the limb muscles and paravertebral muscles.

" if paravertebral muscles are normal, examine two leg muscles innervated by different nerves
" examine muscles innervated by the adjacent roots, above and below, to determine whether it is an isolated or multiple radiculopathy

## C1 RADICULOPATHY

**Etiology**
" extremely rare and probably difficult, if not impossible, to diagnose neurophysiologically
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic neck pain

**Strategy**
" demonstrate neurogenic EMG findings in C1 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG, neurogenic findings in:*
" paravertebral muscles C1

**Expected normal findings**
*EMG*
" paravertebral muscles from C3 downwards

**Procedure**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.sternocleidomastoideus
" m.diaphragma
" m.trapezius
" paravertebral muscles C1-C3

**Revised**
" BF 1996-11-07, 8.5.1997 BF

## C2 RADICULOPATHY

**Etiology**
" extremely rare and probably difficult, if not impossible, to diagnose neurophysiologically
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic neck pain

**Strategy**
" demonstrate neurogenic EMG findings in C2 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG, neurogenic findings in:*
" paravertebral muscles C2

**Expected normal findings**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.sternocleidomastoideus
" m.diaphragma
" m.trapezius
" paravertebral muscles C4

**Procedure**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.sternocleidomastoideus
" m.diaphragma
" m.trapezius
" paravertebral muscles C1-C4

**Revised**
" 8.5.1997 BF, 7.11.1996 BF

## C3 RADICULOPATHY

**Etiology**
" rare and probably difficult to diagnose neurophysiologically
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic neck pain

**Strategy**
" demonstrate neurogenic EMG findings in C3 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG*
" m.sternocleidomastoideus
" paravertebral muscles C3
" m.diaphragma

**Expected normal findings**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.brachioradialis

**Procedure**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.sternocleidomastoideus
" m.trapezius
" paravertebral muscles C2-C4
" *m.diaphragma (optional, examine only if necessary)*

**Modified**
" 8.5.1997 BF, 7.11.1997 BF

## C4 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic neck pain radiating into the shoulder

**Strategy**
" demonstrate neurogenic EMG findings in C4 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG:*
" m.trapezius
" m.sternocleidomastoideus
" m.diaphragma
" paravertebral muscles C4

**Expected normal findings**
*EMG*
" m.rhomboideus
" m.deloideus

**Procedure**
*EMG*
" m.rhomboideus major/m.deltoideus/m.infraspinatus
" m.sternocleidomastoideus
" m.trapezius
" m.triceps
" paravertebral muscles C3-C5
" *m.diaphragma (optional, examine only if necessary)*

**References**

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" Daniels DL, Grogan JP, Johansen JG, Meyer GA, Williams AL, Haughton VM. Cervical radiculopathy: computed tomography and myelography compared. Radiology 1984;151:109-13
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" So YT, Olney RK, Aminoff MJ. A comparison of thermography and electromyography in the diagnosis of cervical radiculopathy. Muscle Nerve 1990;13:1032-6
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**Modified**
" BF 1996-11-07

## C5 RADICULOPATHY

**Etiology**
" herniated intervertebral disc between C4/5
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" 6 % of cervical intervertebral disc herniations occur at this level
" acute, subacute or chronic neck pain radiating into the shoulder
" weakness of upper arm abduction and external rotation
" numbness over the lateral part of the upper arm (not often present)

**Strategy**
" demonstrate neurogenic EMG findings in C5 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG*
" m.rhomboideus major
" m.deltoideus
" m.infraspinatus
" paravertebral muscles C5

**Expected normal findings**
*EMG*
" m.trapezius
" m.brachioradialis
" m.pronator teres
" m.triceps

**Procedure**
*EMG*
" m.rhomboideus major
" m.deltoideus/m.infraspinatus
" m.trapezius
" m.brachioradialis/m.pronator teres
" m.triceps
" paravertebral muscles C4-C6

**References**

" Berger AR, Busis NA, Logigian EL et al: Cervical root stimulation in the diagnosis of radiculopathy. Neurology 1987;37:329-
" Daniels DL, Grogan JP, Johansen JG, Meyer GA, Williams AL, Haughton VM. Cervical radiculopathy: computed tomography and myelography compared. Radiology 1984;151:109-13
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**Modified**
" BF 1996-11-07

## C6 RADICULOPATHY

**Etiology**
" herniated intervertebral disc at the C5/6 level
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic neck pain radiating into the shoulder and arm
" weakness of elbow flexion
" decreased or absent biceps and brachioradialis tendon reflexes
" numbness over the radial side of the forearm and thumb

**Strategy**
" 25 % of cervical intervertebral disc herniations occur at this level
" demonstrate neurogenic EMG findings in C6 innervated muscles
" demonstrate conduction abnormality in the proximal part of the C6 segment
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in:*

" paravertebral muscles C6
" m.biceps brachii
" m.brachioradialis
" m.pronator teres

**Expected normal findings**

*EMG*

" m.rhomboideus
" m.triceps

*Neurography*

" SCS n.medianus to digit I
" SCS n.radialis to digit I
" SCS n.cutaneus antebrachii lateralis

**Procedure**

*EMG*

" m.rhomboideus major
" m.deltoideus/m.infraspinatus
" m.brachioradialis/m.pronator teres
" m.triceps
" m.interosseus dorsalis
" paravertebral muscles C5-C7

*Neurography*

" SCS: n.radialis
" SCS n.medianus
" SCS: n.ulnaris
" CSC n.cutaneus antebrachii lateralis

**References**


" Berger AR, Busis NA, Logigian EL et al: Cervical root stimulation in the diagnosis of radiculopathy. Neurology 1987;37:329-
" Daniels DL, Grogan JP, Johansen JG, Meyer GA, Williams AL, Haughton VM. Cervical radiculopathy: computed tomography and myelography compared. Radiology 1984;151:109-13
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" Massey EW. Hand weakness in elderly patients. Postgrad Med 1989;85:59-70
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**Modified**

" BF 1996-11-07 BF 1997-04-16

## C7 RADICULOPATHY

**Etiology**

" herniated intervertebral disc at the C6/7 level
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" 60 % of cervical intervertebral disc herniations occur at this level
" acute, subacute or chronic neck pain radiating into the shoulder and arm
" weakness of elbow extension and wrist flexion
" decreased or absent triceps tendon reflex
" numbness over the forefinger and middle finger

**Strategy**

" demonstrate neurogenic EMG findings in C7 innervated muscles
" demonstrate conduction abnormality in the proximal part of the C7 segment
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in:*

" m.triceps brachii
" m.flexor carpi radialis
" m.latissimus dorsi/m.pectoralis major
" m.extensor digitorum communis
" paravertebral muscles C7

*Neurography*
" H-reflex to m.flexor carpi radialis

**Expected normal findings**

*EMG*
" m.biceps brachii
" m.abductor digiti minimi/m.interosseus dorsalis I

*Neurography*
" SCS n.medianus dig II and III

**Procedure**

*EMG*
" m.biceps/m.brachioradialis
" m.flexor carpi radialis
" m.triceps
" m.interosseus dorsalis I
" m.abductor pollicis brevis
" paravertebral muscles C6-C8

*Neurography*
" SCS: n.radialis
" SCS n.medianus
" SCS: n.ulnaris
" MCS: n.medianus: H-reflex to m.flexor carpi radialis

**References**


" Berger AR, Busis NA, Logigian EL et al: Cervical root stimulation in the diagnosis of radiculopathy. Neurology 1987;37:329-
" Daniels DL, Grogan JP, Johansen JG, Meyer GA, Williams AL, Haughton VM. Cervical radiculopathy: computed tomography and myelography compared. Radiology 1984;151:109-13
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**Modified**

" BF 1996-11-07, 16.4.1997 BF

## C8 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" acute, subacute or chronic neck pain radiating into the shoulder and arm
" weakness of the intrinsic hand muscles
" numbness of the index and little fingers
" may be accompanied by Horner's syndrome

**Strategy**

" demonstrate neurogenic EMG findings in C8 innervated muscles
" demonstrate conduction abnormality in the proximal part of the C8 segment
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in:*
" m.extensor indicis proprius
" m.interosseus dorsalis I/abductor digit minimi
" m.abductor pollicis brevis
" paravertebral muscles C8

**Expected normal findings**

*EMG*
" m.pronator teres
" m.triceps

*Neurography*
" SCS from dig V normal

**Procedure**

*EMG*

" m.brachioradialis/m.pronator teres
" m.triceps
" flexor carpi radialis
" m.interosseus dorsalis I
" m.extensor indicis
" m.opponens pollicis/m.abductor pollicis brevis
" paravertebral muscles C7-Th1

*Neurography*

" SCS: n.radialis
" SCS n.medianus
" SCS: n.ulnaris

**References**


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**Modified**

" BF 1996-11-07, 16.4.1997

## TH1 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" acute, subacute or chronic neck pain radiating into the shoulder arm
" weakness of thumb
" numbness over the ulnar side of the forearm

**Strategy**

" demonstrate neurogenic EMG findings in Th1 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in:*

" m.opponens pollicis/m.abductor pollicis brevis
" m.interosseus dorsalis
" paravertebral muscles Th1

**Expected normal findings**

*EMG*

" m.triceps/m.extensor digitorum communis

*Neurography*

" SCS normal in cutaneus antebrachii medialis

**Procedure**

*EMG*

" m.brachioradialis/m.pronator teres
" m.triceps
" m.interosseus dorsalis I
" m.extensor indicis
" m.opponens pollicis/m.abductor pollicis brevis
" paravertebral muscles C8-Th1

*Neurography*

" SCS: n.radialis
" SCS n.medianus
" SCS: n.ulnaris

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**Modified**
" BF 1996-11-07, 16.4.1997

## TH2-TH10 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic pain radiating unilaterally over the of thorax

**Strategy**
" demonstrate neurogenic EMG findings in paravertebral muscles and intercostal muscles at the respective level

**Expected abnormal findings**
*EMG, neurogenic findings in:*
" appropriate paravertebral muscles
" appropriate intercostal muscles
" at levels Th7-Th12 m.rectus abdominis

**Procedure**
*EMG*
" appropriate paravertebral muscles
" appropriate intercostal muscles
" m.rectus abdominis, the upper part is innervated from Th7 to Th9 and the lower part from Th9 to Th12

**Modified**
" BF 1996-11-07

## TH11 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" pain radiating unilaterally over the abdomen

**Strategy**
" demonstrate neurogenic EMG findings in Th11 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**
*EMG, neurogenic findings in*
" paravertebral muscles Th11
" m.rectus abdominis pars inferior

**Procedure**
*EMG*
" paravertebral muscles Th10-12
" m.rectus abdominis

**Modified**
" 8.5.1997 BF, 7.11.1996 BF

## TH12 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Strategy**

" demonstrate neurogenic EMG findings in Th11 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in*
" paravertebral muscles Th12
" m.rectus abdominis pars inferior

**Expected normal findings**

*EMG*
" m.iliopsoas

**Procedure**

*EMG*
" paravertebral muscles Th11-L1
" m.rectus abdominis pars inferior
" m.transversus abdominis
" m.iliopsoas

**References**

**Modified**
" BF 1996-11-07

## L1 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic pain radiating unilaterally over the abdomen

**Strategy**
" demonstrate neurogenic EMG findings in L1 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic in*
" m.rectus abdominis pars inferior
" paravertebral muscles L1

**Expected normal findings**

*EMG*
" m.adductor magnus

**Procedure**

*EMG*
" paravertebral muscles Th12-L2
" m.rectus abdominis
" m.iliopsoas
" m.adductor magnus

**References**
"

**Modified**
" BF 1996-11-07

## L2 RADICULOPATHY

**Etiology**
" herniated intervertebral disc
" spondylarthrosis
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**
" acute, subacute or chronic pain radiating unilaterally into the thigh
" weakness of the thigh flexion
" numbness over the anterior part of the thigh

**Strategy**
" demonstrate neurogenic EMG findings in L2 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic in:*
" m.iliopsoas
" m.adductor magnus
" m.vastus lateralis/m.vastus medialis
" paravertebral muscles L2

**Expected normal findings**

*EMG*
" m.tibialis anterior

*Neurography*
" saphenus SCS normal

**Procedure**

*EMG*
" paravertebral muscles L1-L3
" m.iliopsoas

" m.adductor magnus
" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior

*Neurography (optional)*

" SCS n.saphenus

**References**

"

**Revised**

" 8.5.1997 BF

## L3 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" spinal anesthesia
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" acute, subacute or chronic pain radiating unilaterally into the thigh
" weakness of the thigh flexion and knee extension
" numbness over the anterior part of the thigh and knee

**Strategy**

" demonstrate neurogenic EMG findings in L3 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG neurogenic in:*

" m.adductor magnus
" m.iliopsoas
" m.vastus lateralis/m.vastus medialis
" paravertebral muscles L3

**Expected normal findings**

*EMG*

" m.tibialis anterior

*Neurography*

" saphenus SCS normal

**Procedure**

*EMG*

" paravertebral muscles L2-L4
" m.iliopsoas
" m.adductor magnus
" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior

*Neurography (optional)*

" SCS n.saphenus

**References**

"

**Modified**

" 8.5.1997 BF, 7.11.1996 BF

## L4 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" spinal anesthesia
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" acute, subacute or chronic back pain radiating into the leg
" numbness of the anterior part of the leg
" loss of patellar reflex
" weakness of knee extension

**Strategy**

" demonstrate neurogenic EMG findings in L4 innervated muscles
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG neurogenic in*

" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior
" paravertebral muscles L4

**Expected normal findings**

*EMG*

" m.peroneus longus/m.extensor hallucis longus

*Neurography*

" SCS n. saphenus

**Procedure**

*EMG*

" paravertebral muscles L3-L5
" m.iliopsoas
" m.adductor magnus
" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior
" m.peroneus longus/m.extensor hallucis longus
" m.gastrocnemius caput mediale/m.gastrocnemius caput laterale

*Neurography*

" SCS: n.saphenus
" MCS: n.peroneus

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## L5 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" spinal anesthesia
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" acute, subacute or chronic back pain radiating into the leg
" numbness over the dorsal side of the foot
" decreased toe and ankle dorsal flexion

**Strategy**

" demonstrate neurogenic EMG findings in L5 innervated muscles
" demonstrate conduction abnormality in the proximal part of the L5 segment
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic*

" paravertebral muscles L5
" peroneus longus/extensor hallucis longus/tibialis anterior/flexor hallucis longus/ tibialis posterior
" tensor fascia latae/semitendinosus

*Neurography*

" M-wave amplitude may be low in m.extensor digitorum brevis and other muscles innervated by n.peroneus
" F-waves in peroneal nerve may be abnormal

**Expected normal findings**

*EMG*

" vastus lateralis/vastus medialis
" m.gastrocnemius caput mediale/laterale
" m.gluteus maximus/m.biceps femoris

*Neurography*

" SCS normal: suralis/peroneus superficialis
" H-reflex normal in calf muscles

**Procedure**

*EMG*

" paravertebral muscles L4-S1
" m.vastus lateralis/m.vastus medialis (L4)
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus (distal L5)
" m.tensor fascia latae/m.gluteus medius/m.semitendinosus (proximal L5)
" m.gluteus maximus/m.biceps femoris (proximal S1)
" m.gastrocnemius caput mediale/m.gastrocnemius caput laterale (distal S1)

*Neurography*

" SCS: n.suralis
" MCS: n.peroneus
" MCS: n.tibialis
" MCS: n.tibialis H-reflex to m.soleus

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## S1 RADICULOPATHY

**Etiology**

" herniated intervertebral disc
" spondylarthrosis
" spinal anesthesia
" very rarely neuroma, unless the patient has Mb von Recklinghausen

**Clinical features**

" usually acute or subacute lumbosacral pain radiating in the leg
" numbness on the lateral side of the foot
" Achille's reflex reduced or absent

**Strategy**

" demonstrate neurogenic EMG findings in S1 innervated muscles
" demonstrate conduction abnormality in the proximal part of the S1 segment
" examine muscles innervated by the adjacent roots

**Expected abnormal findings**

*EMG, neurogenic findings in*
" paravertebral muscles S1
" m.gluteus maximus/m.biceps femoris caput brevis
" m.gastrocnemius caput mediale/m.gastrocnemius caput laterale/m.soleus

*Neurography*
" H-reflex abnormal
" F-waves to calf muscles abnormal

**Expected normal findings**

*EMG*
" peroneus longus/flexor hallucis longus/extensor hallucis longus
" tensor fascia latae

*Neurography*
" n.suralis

**Procedure**

*EMG*
" paravertebral muscles L5-S1
" m.vastus lateralis/m.vastus medialis (L4)
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus (distal L5)
" m.tensor fascia latae/m.gluteus medius/m.semitendinosus (proximal L5)
" m.gluteus maximus/m.biceps femoris (proximal S1)
" m.gastrocnemius caput mediale/m.gastrocnemius caput laterale (distal S1)

*Neurography*
" SCS: n.suralis
" MCS: n.peroneus
" MCS: n.tibialis
" MCS: n.tibialis H-reflex to m.soleus

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## LOW BACK PAIN WITH NO RADICULOPATHY

**Strategy**

" normal findings in clinically suspected roots.
" usually it is sufficient to examine L4, L5 and S1 roots.
" if there are clinically symptoms or signs of L1-L3 involvement; check also those.

**Expected normal findings**

*EMG in one muscle from each of the following five groups and the corresponding paravertebral muscles*
" L4: m.vastus lateralis/m.vastus medialis
" L5, proximal muscles. m.tensor fascia latae/m.gluteus medius/m.semitendinosus/m.semimembranosus

" L5, distal muscles: m.peroneus longus/m.extensor hallucis longus/m.tibialis anterior or m.flexor hallucis longus
" S1, proximal muscles. m.gluteus maximus/m.biceps femoris caput brevis
" S1, distal muscles: m.gastrocnemius caput laterale/m.gastrocnemius caput mediale/m.soleus

*Neurography*

" H-wave from soleus
" n.tibialis F-wave
" n.peroneus F-wave

**Procedure**

*EMG*

" paravertebral muscles
" m.vastus lateralis/m.vastus medialis (L4)
" m.tibialis anterior/m.peroneus longus/m.extensor hallucis longus (distal L5)
" m.tensor fascia latae/m.gluteus medius/m.semitendinosus (proximal L5)
" m.gluteus maximus/m.biceps femoris (proximal S1)
" m.gastrocnemius caput mediale/m.gastrocnemius caput laterale (distal S1)

*Neurography*

" SCS: n.saphenus
" SCS: n.peroneus superficialis
" SCS: n.suralis
" MCS: n.peroneus
" MCS: n.tibialis
" MCS: n.tibialis H-reflex to m.soleus

### NECK PAIN WITH NO RADICULOPATHY

**Strategy**

" normal findings in clinically suspected roots. Usually it is sufficient to examine C5, C6, C7, C8 and Th1 roots.
" if there are signs of involvement in neighboring areas check also those.

**Expected normal findings**

*EMG in one muscle from each of the following five groups and the corresponding paravertebral muscles*

" m.rhomboideus/m.deltoideus
" m.biceps brachii/m.brachioradialis
" m.triceps brachii/m.flexor carpi radialis
" m.extensor indicis proprius/m.interosseus dorsalis I
" m.abductor pollicis brevis/m.interosseus dorsalis I
" paravertebral muscles C5-Th1

*Neurography*

" H-wave from m.flexor carpi radialis
" n.medianus and n.ulnaris F-waves
" SCS: n.ulnaris and n.medianus

**Procedure**

*EMG*

" M.delotideus/m.infraspinatus (C5)
" m.biceps/m.brachioradialis/m.pronator teres (C6)
" m.triceps /m.flexor carpi radialis (C7)
" m.interosseus dorsalis (C8)
" m.opponens pollicis/m.abductor pollicis brevis (Th1)
" paravertebral muscles

*Neurography*

" SCS: n.radialis
" SCS n.medianus
" SCS: n.ulnaris
" MCS: n.medianus: H-reflex to m.flexor carpi radialis

## 7. DISORDERS OF THE SPINAL CHORD AND CENTRAL NERVOUS SYSTEM WITH NEUROMUSCULAR ABNORMALITIES

### CERVICAL SYRINGOMYELIA

**Etiology**

" syringomyelia is a chronic disorder involving the spinal cord or medulla or both.
" it is characterized by the development of cavitation and gliosis within the spinal chord or medulla
" intraspinal dilatation of cavity is due to developmental, vascular or traumatic causes

**Clinical features**

" onset usually between 20 and 40 years, but may be seen both in younger and older age groups
" dissociated sensory loss; loss of pain and temperature, but preservation of touch
" muscle weakness and atrophy
" impairment of long tract functions
" syringomyelia is considered slowly progressive and degenerative, but there are indications that progression may be arrested by appropriate treatment.

**Strategy**

" demonstrate neurogenic abnormalities in muscles innervated from affected myotomes
" exclude peripheral nerve lesions (ulnar nerve lesions, lower part of plexus and C8 radiculopathy)
" differentiate from monomelic spina muscular atrophy, motor neuropathy with multifocal conduction block and ALS

**Expected abnormal findings**

*EMG*

" m.interossseus dorsalis/m.abductor digiti minimi
" m.flexor carpi ulnaris
" m.extensor indicis/m.extensor digitorum communis

*Neurography*

" MCS, AMPL may be reduced, in the presence of low AMPL CV may be reduced

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*Neurography (bilaterally)*

" MCS n.medianus, n.ulnaris (fractionated across the elbow)
" SCS n.ulnaris, n.medianus, n.radialis, n.cutaneus antebrachii lateralis, n.cutaneus antebrachii medialis

*EMG (bilaterally)*

" m.interosseus dorsalis I (n.ulnaris)
" m.opponens pollicis/m.abductor pollicis brevis (n.medianus)
" m.extensor indicis proprius (C8 and inferior trunk)
" m.triceps brachii
" m.biceps brachii/m.deltoideus
" m.trapezius (to evaluate brainstem involvement, syringobulbia)
" m.hyoglossus/m.hyoglossus (to evaluate brainstem involvement, syringobulbia)

*Evoked potentials*

" MEP and SEP may be helpful in the evaluation of long tract involvement

**References**

"

**Revised**

"

## FRIEDREICH'S ATAXIA

**Etiology**

" autosomal recessive inheritance
" gene location chromosome 9cen-q21
" the mutation consists of an unstable expansion of GAA repeats in the first intron of the frataxin gene on chromosome 9, which encodes a protein of unknown function

**Clinical features**

" prevalence 1-2 per 100 000
" onset of symptoms usually between 8-15 (range 2-50 years)
" staggering gait often first symptom, sometimes clumsiness
" ataxia
" loss of tendon reflexes, sometimes preserved quite late
" dysarthria
" scoliosis is common
" high foot arches
" eventually the patients become bedridden
" decreased longevity

**Strategy**

" demonstrate abnormal SCS amplitude, CV is normal or only slightly reduced
" EMG and MCS normal
" differentiate from polyneuropathy

**Expected abnormal findings**

*Neurography*

" SCS: reduced SCS amplitude

**Expected normal findings**

*EMG*

*Neurography*

" MCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis, n.medianus and n.ulnaris bilaterally
" MCS: n.peroneus and n.medianus on one side

**References**

" Harding AE: Friedreich's ataxia: a clinical and genetic study of 90 families with an analysis of early diagnostic criteria and intrafamilial clustering of clinical features. Brain 1981;104:589-597
" Hammans SR. The inherited ataxias and the new genetics. J Neurol Neurosurg Psychiat 1996;61:327-332.

**Modified**

" October 21st 1996, BF

## MACHADO-JOSEPH DISEASE,

**Etiology**

" autosomal dominant inheritance
" gene maps to long arm of chromosome 14q24.3-32.1
" the mutation consists of an unstable expansion of CAG repeats in the 3´end of the coding region
" the variability of the clinical severity depends on the number of trinucleotide repeats

**Clinical features**

" Four subtypes have been suggested:
" I. The age of onset is in the mid-20s. It is noted for its relative lack of cerebellar findings. This syndrome is characterized by the pyramidal and extrapyramidal signs of spasticity, hyperreflexia, Babinski's sign, clonus, rigidity, dystonia. There is also the onset of facial and lingual fasciculations, nystagmus, and PEO.
" II. Onset in the fourth through sixth decades. The second subtype includes primarily cerebellar and extrapyramidal features. There is truncal ataxia in association with a lurching gait. There is progression to dysarthria and the appearance of dystonia and occasionally chorea.
" III: The onset here is in the fifth through seventh decades. The third subtype is characterized by the cerebellar signs in association with a distal polyneuropathy. There is distal sensory loss and amyotrophy with hyporeflexia.
" IV. The fourth subtype is dominated by neuropathy and parkinsonian-like features.
" The diagnosis is made by the positive family history, the cerebellar syndrome, and the pyramidal and extrapyramidal findings. These in combination with bulging eyes, lid retraction, and the PEO are unique to this syndrome.

**Strategy**

" demonstrate abnormal findings in sensory nerves
" muscles show neuropathic changes in many patients, especially in the later stages
" CNS also affected

**Expected abnormal findings**

*Neurography*

" SCS: reduced or absents SCS responses
" neuropathic EMG abnormalities with fasciculations
" abnormal VEP, SEP and BAEP

**Expected normal findings**

*EMG*

*Neurography*

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis, n.medianus and n.ulnaris bilaterally
" MCS: n.peroneus and n.medianus on one side

*Evoked potentials*

" BAEP
" SEP
" VEP

**References**

" Colding-Jorgensen E, Sorensen SA, Asholdt L, Lauritzen M. Electrophysiological findings in a Danish family with Machado Joseph disease. Muscle Nerve 1996; 19:743-750.
" Hammans SR. The inherited ataxias and the new genetics. J Neurol Neurosurg Psychiat 1996;61:327-332.
" Rosenberg RN: Joseph disease: an autosomal dominant motor system degeneration. In Duvoisin RC, Plaitakis A (eds): The Olivopontocerebellar Atrophies, New York, Raven Press, 1984

**Modified**

" October 21st 1996, BF

## METACHROMATIC LEUKODYSTROPHY

**Etiology**

" deficiency of arylsuphatase a
" autosomal recessive inheritance
" linked to chromosome 22q-22ter13
" the metachromatic leukodystrophies are a group of genetically determined disorders affecting the nervous system
" CNS and PNS is affected
" accumulation of galactosyl sulfatide and other sulfatide lipids

**Clinical features**

" late infantile form:
    " onset between1-4 years
    " initial symptom walking difficulty
    " mental retardation
    " later ataxia an tetraplegia
    " blindness and deafness
" Juvenile and adult onset form
    " similar to infantile form but later onset

**Strategy**

" in late infantile form demonstrate demyelinating sensory motor polyneuropathy
" in juvenile adult form the demyelinating polyneuropathy is not as prominent

**Expected abnormal findings**

*Neurography*

" in the late infantile form reduced CV, often in the range of 10-20 m/s, SCS absent sensory responses reduced in amplitude or absent
" in the juvenile adult form the CV is slightly reduced

*EMG*

" neurogenic abnormalities, especially in distal muscles

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis bilaterally
" MCS: n.peroneus and n.medianus bilaterally

**References**

" Guffon N, Souillet G, Maire I, et al. Juvenile metachromatic leukodystrophy: neurological outcome two years after bone marrow transplantation. J Inherit Metab Dis 1995;18:159-161
" Aicardi J. The inherited leukodystrophies: a clinical overview. J Inherit Metab Dis 1993;16:733-43.
" Clark JR, Miller RG, Vidgoff JM. Juvenile-onset leukodystrophy: biochemical and electrophysiological study. Neurology 1979;29:346-53.

**Modified**

" 25.3.1997 BF.

## GALACTOSYLCERAMIDE LIPIDOSIS (KRABBE DISEASE, GLOBOID CELL LEUKODYSTORPHY)

**Etiology**

" the gene has been mapped to chromosome 14q24-q32
" galactocylcereamide β-galactosidase deficiency
" autosomal recessive inheritance

**Clinical features**

" normal neonatal development
" onset at the age of 3 to 6 months (cases with later onset and slower progression have been described)
" mental retardation
" hypertonicity of limbs
" aggravated reflexes
" children become blind and deaf
" CSF protein elevated
" death by 2 years of age
" prenatal diagnosis is available

**Strategy**

" demonstrate sever demyelinating polyneuropathy

**Expected abnormal findings**

*Neurography*

" SCS: CV reduced by 50 %, reduced SCS amplitude
" MCS: CV reduced by 50 %

*EMG*

" Neurogenic abnormalities

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis bilaterally
" MCS: n.peroneus and n.medianus bilaterally

*Evoked potentials*

" BAEP is often abnormal, I-III interpeak latency prolonged

**References**

" Rafi MA, Luzi P, Chen YQ, Wenger DA. A large deletion together with a point mutation in the GALC gene is a common mutant allele in patients with infantile Krabbe disease. Hum Molec Genet 1995;4:1285-1289.
" Oehlmann R, Zlotogora J, Wenger DA, Knowlton RG. Localization of the Krabbe disease gene (GALC) on chromosome 14 by multipoint linkage analysis. Am J Hum Genet 1993;53:1250-1255.
" Hagberg B. Krabbe's disease: clinical presentation of neurological variants. Neuropediatrics 1984;15:11-15.

**Modified**

" 26.3.1997 BF

## ADRENOLEUKODYSTROPHY/ADRENOMYELONEUROPATHY

**Etiology**

" x-linked recessive inheritance
" deficient β-oxygenation of very long chain fatty acids
" accumulation of very long chain (25-26 carbon chain fatty acids)

**Clinical features**

" onset of symptoms varies markedly, may begin in childhood or adulthood
" progressive cerebral degeneration
" cerebellar ataxia, pyramidal tract symptoms
" cortical blindness, deafness, spasticity, seizures
" peripheral neuropathy
" sometimes adrenal failure

**Strategy**

" demonstrate demyelinating polyneuropathy

**Expected abnormal findings**

*EMG*

" mild neurogenic abnormalities

*Neurography*

" SCS: reduced CV and amplitude

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis bilaterally
" MCS: n.peroneus and n.medianus bilaterally

**References**

" Fournier B, Smeitink JA, Dorland L, Berger R, Saudubray JM, Poll-The BT. Peroxisomal disorders: a review. J Inherit Metab Dis 1994;17:470-86.
" Verma NP, Hart ZH, Nigro M. Electrophysiological studies in neonatal adrenoleukodystrophy. Electroenceph Clin Neurophysiol 1985;60:7-15.

**Modified**

" 25.3.1997 BF

# 8. MISCELLANEOUS

## BENIGN FASCICULATION

**Etiology**

" unknown
" sporadic

**Clinical features**

" fasciculations in muscles
" onset may be at any age
" most persons seeking medical advice for this disorder are medical personnel, especially medical students

**Strategy**

" demonstrate fasciculations in muscles but no signs of denervation or reinnervation
" differentiate from ALS, MNM, SMA and other neurogenic disorders

**Expected abnormal findings**

*EMG*

" fasciculations

**Expected normal findings**

*EMG*

" no fibrillations or positive sharp waves
" MUP analysis

*Neurography*

" MCS
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus and n.medianus on one side

**Referrences**

" Blexrud MD, Windebank AJ, Daube JR. Long term follow-up of 121 patients with benign fasciculations. Ann Neurol 1993; 43:622-625
" Reed DM, Kurland LK. Muscle fasciculations in a healthy population. Arch Neurol 1963; 9:363-367

**Modified**

" 25.3.1997 BF

## NEUROMYOTONIA (ISAAC'S SYNDROME, CONTINOUS MUSCLE FIBER ACTIVITY)

**Etiology**

" probably autoimmune disorder
" antibodies against voltage gated potassium channels
" often occurs sporadically without an apparent cause
" may be caused by penicillamine
" sometimes a paraneoplastic feature, especially related with lung cancer or thymoma, plasmacytoma and IgM paraproteinemia

**Clinical features**

" muscle stiffness and rippling of the muscles

" usually responds well to phenytoin or carbamazapine, plasmapheresis is also effective

**Strategy**

" demonstrate neuromyotonic activity
" differentiate from myotonia

**Expected abnormal findings**

*EMG*

" abundant neuromyotonia, especially in distal muscles

*Neurography*

" MCS may sometimes be abnormal, reduced CV and amplitude

**Expected normal findings**

*EMG*

" MUP analysis normal

*Neurography*

" MCS, sometimes abnormal
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus, n.tibialis, n.ulnaris and n.medianus on one side

**References**

" Layzer RB. Neuromyotonia: a new autoimmune disease (editorial). Ann-Neurol. 1995;38:701-702
" Partanen VS, Soininen H, Saksa M, Riekkinen P. Electromyographic and nerve conduction findings in a patient with neuromyotonia, normocalcemic tetany and small-cell lung cancer. Acta-Neurol-Scand. 1980; 4:216-26
" Walsh JC. Neuromyotonia: an unusual presentation of intrathoracic malignancy. J Neurol Neurosurg Psychiatry 1976;11:1086-1091
" Brown TJ. Isaacs syndrome. Arch Phys Med Rehabil 1984;65:27-29
" Newsom-Davis J, Mills KR. Immunological associations of acquired neuromyotonia (Isaacs' syndrome). Report of five cases and literature review. Brain 1993;116:453-69
" Hart IK, Waters C, Vincent A, Newland C, Beeson D, Pongs O, Morris C, Newsom-Davis J. Autoantibodies detected to expressed K+ channels are implicated in neuromyotonia. Ann Neurol 1997; 41: 238-246

**Modified**

" 26.3.1997 BF

## STIFF MAN SYNDROME

**Etiology**

" probably autoimmune disorder
" antibodies against glutamic acid decarboxylase (GAD)

**Clinical features**

" onset age 8-76, average around 40 years
" men and women equally affected
" initially tightness and aching of axial muscles
" progression to symmetric stiffness of trunk and limb muscles
" sudden stimuli, auditory and sensory but not visual, painful muscle spasms (spasmodic myoclonia)
" hyperlordosis and elevation of shoulders
" muscles stiffness disappears during sleep and with benzodiazepine treatment
" tendon reflexes normal or increased
" often associated with type 1 diabetes (30-50% of the patients) and other immune mediated disorders (myasthenia gravis, vitiligo, pernicious anemia, thyroiditis, etc.)
" antibodies against GAD

**Strategy**

" demonstrate abundant ongoing EMG activity in axial muscles and limb muscles
" differentiate from myotonia

**Expected abnormal findings**

*EMG*

" abundant EMG activity, especially in axial muscles

**Expected normal findings**

*EMG*

" normal insertional activity, if it can be evaluated
" MUP analysis normal

*Neurography*

" MCS
" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor pollicis brevis/m.extensor digitorum communis/m.flexor carpii radialis
" m.vastus lateralis/m.vastus medialis/m.tensor fascia latae
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus, n.tibialis, n.ulnaris and n.medianus on one side

*Effect of benzodiazepines*

" monitor the effect of IV injection of diazepam with EMG, in stiff man syndrome the EMG activity should be considerably reduced following the injection

**Note**

" abnormal long loop reflexes have been described in stiff man syndrome
" abnormal flexor reflexes have been described in stiff man syndrome

**References**


" Auger RG: Diseases associated with excess motor unit activity. Muscle Nerve 1994; 17:1250-1263.
" Gordon E, Januszko M, Kaufman L: A critical survey of stiff-man syndrome. Am J Med 1967; 42:582-599.
" McEvoy K: Stiff-Man syndrome. Seminars in Neurology 1991; 11:197-205.
" Meink H, Ricker K, Hülser P, Solimena M: Stiff man syndrome: neurophysiological findings in eight patients. J Neurol 1995; 242:134-142.
" Moersh F, Woltman H: Progressive fluctuating muscular rigidity and spasms ("stiff-man syndrome"): Report of a case and some observations on 13 other cases. Mayo Clin Proc 1956; 31:421-427.


**Modified**

" 6.11.1996 BF, 26.3.1997 BF

## TETANUS

**Etiology**

" toxin produced by Clostridium tetani under anaerobic conditions
" toxin transported after endocytosis by retrograde transport to the spinal chord
" inhibits release of glycine and GABA
" both $\alpha$ and $\gamma$ motoneurones are disinhibited
" in high concentrations the effects of tetanus toxcin are similar to botulinus toxin

**Clinical features**

" occurs very rarely because of effective immunization
" may occur in patints with proper immunization!
" presents 3 to 20 days following exposure to bacteria
" localized type: painful spasms near injury, limited spasms last for two weeks
" generalized type is more common: trismus, opistotonus, reflex spasms, stiffness, neck rigidity, dysphagia: tetanic spasms
" autonomic over activity: hypertension, dysrythmias
" in long standing cases there is denervation of muscles
" diagnosis is clinical there are no specific tests

**Strategy**

" demonstrate abundant EMG activity in axial muscles and limb muscles
" differentiate from stiff man syndrome and epilepsy

**Expected abnormal findings**

*EMG*

" abundant EMG activity, especially in axial muscles
" after one week signs of acute denervation on EMG

*Neurography*

" initially normal findings
" after one week reduced MCS amplitude

**Expected normal findings**

*Neurography*

" SCS

**Procedure**

*EMG*

" m.deltoideus/m.biceps brachii
" m.interosseus dorsalis I/m.abductor digiti minimi/m.abductor pollicis
" m.vastus lateralis/m.vastus medialis
" m.tibialis anterior

*Neurography*

" SCS: n.suralis and n.radialis on one side
" MCS: n.peroneus, n.tibialis, n.ulnaris and n.medianus on one side

**References**


" Benson CA, Harris AA: Acute neurologic infections. Med Clin North Am 1986.70:987
" Kaeser HE, Muller Müller HR, Friedrich B: The nature of tetraplegia in infectious tetanus. Eur Neurol 1968;1:17
" Crone NE, Reder AT. Severe tetanus in immunized patients with high anti-tetanus titers. Neurology. 1992;42:761-764
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**Modified**

" 16.4.1997 BF