

Congenital Myasthenic Syndromes

- Clinical Data -

SAMPLES FOR MOLECULAR GENETIC TESTINGS

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DNA / 10 ml EDTA-Blood

to

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included are samples of

patient

name:

date of birth:

female

male

relatives:
.....
.....
.....

if possible, please send samples of affected and / or unaffected relatives...

sample sent by :

institute:

supervising
physician:

telephone/e-mail

CLINICAL DATA

1. FAMILY HISTORY

are there affected relatives?

no information

no

yes ...if yes, who is affected?

.....
.....

how many healthy siblings has the patient?

.....
.....

origin of the family?

.....
.....

consanguinity in the family?

no information

no

yes ...if yes, please explain / show pedigree

.....
.....

2. HISTORY

onset of disease: at birth
 first year of life
 childhood **at age:** _____
 adolescence **at age:** _____
 adulthood **at age:** _____

motor milestones: normal
 delayed
ability to walk at age: _____

course of disease:
 progressive
 no / slight progression
 tendency towards remission

episodic crisis
 no
 yes → sudden severe weakness
 respiratory insufficiency
 precipitated by: _____
 frequency/age: _____

muscle biopsy performed?
 no yes, result: _____

3. CLINICAL SYMPTOMS

weakness:
 generalized
 selected muscle groups → ocular muscles
 others:
 exercise induced
 constant
 moderate weakness
 severe weakness → ambulance not restricted
 restricted: walking distance: _____
 loss of ambulance / wheelchair

Involved muscle groups:
 ocular muscles → ptosis
 extraocular muscles / limited eye movement
 bulbar muscles → facial weakness
 nasal speech
 swallowing difficulties
 chewing difficulties
 extremities distal = proximal
 predominantly proximal muscle groups involved
 predominantly distal muscle groups involved

limb girdle scapulae alatae
 waddling gait
 axial muscle involvement (scoliosis ?)
 respiratory muscles

muscular atrophy: no yes

abnormal tendon reflexes: no yes

4. SPECIALS

slow pupillary light response: no yes:
selectively severe weakness of cervical, wrist and finger extensor muscles:
 no yes:
congenital contractures: no yes:
congenital malformations: no yes:
others: _____

5. ELEKTROPHYSIOLOGY

EMG: not done normal
 neuropathic changes myopathic changes
 others: _____

decremental response:
 distal muscle groups proximal muscle groups
 yes (%) yes (%)
 no no
 not determined not determined

facial muscles **response to single nerve stimuli:**
 yes (%) single CMAP response
 no double CMAP response
 not determined not determined

... further studies

single fiber EMG _____
nerve conduction _____

6. SERUM-CREATINEKINASE

no info
 normal
 elevated to _____

7. ANTI-ACETYLCHOLINE RECEPTOR ANTIBODIES

not present
 present
 not determined

8. ACETYLCHOLINESTERASE INHIBITORS:

positive effect
 no clear effect
 negative effect
 not done

9. FURTHER THERAPY:

immunosuppressive treatment (steroids, immunoglobulins, etc....)

not done
 unsuccessful

Successful treatment with

Thank you !