

Congenital Myasthenic Syndromes - Clinical Data -

SAMPLES FOR MOLECULAR GENETIC TESTINGS

Contact:

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DNA / 10 ml EDTA-blood

to

**Labor für Molekulare Myologie
AG Prof. Lochmüller
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D-81377 München**

samples of

patient

name:

date of birth:

female

male

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

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

REPORT TO/sample sent by:

institute (please indicate complete postal address including area code):

supervising physician:

e-mail and telephone

CLINICAL DATA

1. FAMILY HISTORY

are there affected relatives?

no information

no

yes ...if yes, who is affected?

.....

how many healthy siblings does the patient have?

.....

ethnic 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
- exacerbations / episodic crisis**
- no
 - yes → sudden severe weakness
 - with respiratory insufficiency
 - precipitated by:
 - frequency/age:

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/dysmorphisms:** no yes
- please specify:**

5. ELECTROPHYSIOLOGY

- EMG:** not done normal
- neuropathic changes
 - myopathic changes
 - others:
- decremental response:**
- distal muscle groups proximal muscle groups
 - yes (%)
 - no
 - not determined
- facial muscles **response to single nerve stimuli:**
- yes (%)
 - no
 - not determined
 - single CMAP response
 - double CMAP response
 - not determined
- ... further studies**
- single fiber EMG nerve conduction

6. SERUM-CREATINEKINASE/MUSCLE BIOPSY

- | | |
|--|---|
| CK-level | Muscle biopsy |
| <input type="checkbox"/> no info | <input type="checkbox"/> not done |
| <input type="checkbox"/> normal | <input type="checkbox"/> normal |
| <input type="checkbox"/> mildly elevated < 5x normal | <input type="checkbox"/> type II atrophy |
| <input type="checkbox"/> elevated > 5x normal | <input type="checkbox"/> tubular aggregates |

7. ANTI-ACETYLCHOLINE RECEPTOR/ANTI-MuSK ANTIBODIES

- | | |
|---|---|
| anti-AChR | anti-MuSK |
| <input type="checkbox"/> not present | <input type="checkbox"/> not present |
| <input type="checkbox"/> present | <input type="checkbox"/> present |
| <input type="checkbox"/> not determined | <input type="checkbox"/> not determined |

8. THERAPY:

- intravenous AChE-inhibitors (e.g. tensilon)**
- not done
 - no or no clear effect
 - positive effect
 - worsening of symptoms
- AChE-inhibitors (orally, e.g. mestinone)**
- no treatment
 - no or no clear effect
 - positive effect
 - worsening of symptoms
- 3,4-Diaminopyridine**
- no treatment
 - no effect
 - successful

other therapeutic trials (e.g. ephedrine fluoxetine, quinidine)?:

- immunosuppressive treatment (steroids, immunoglobulins, etc....)**
- not done
 - unsuccessful

Thank you !