Diagnostic and Advisory Service for Rare Neuromuscular Diseases NHS

Oxford - Referral Centre for Congenital Myasthenic Syndromes

Funded through the National Specialist Commissioning (NSC), Department of Health

Pre-Referral Form

Note: We cannot accept samples unless this is completed.

Currently a free service to patients living in England & Scotland.
Charges for DNA samples from elsewhere will be made to the relevant Health Authority (please enquire).

CMS comprises a heterogeneous group of conditions: to date 11 genes comprising >200 different mutations have been identified. **Because of this it is preferred that we see your patient in the clinic to help direct the genetic screening appropriately.**
The results and advice we can offer include:

- clinical assessment, treatment advice and education to the patient and their family.
- specialised electromyography
- analysis of DNA from a blood sample
- antibody (anti-acetylcholine receptor and anti-MuSK) assays
- pre-natal screening

Two levels of service are offered please indicate which you require below:

☐ **(Preferred)** Clinical assessment, specialised investigations and DNA analysis on blood sample.
Return FORM ONLY to: NCG Administrator, Department of Clinical Neurology, Level 3
West Wing, John Radcliffe Hospital, Headington, Oxford. OX3 9DU
Tel: 01865 231897 Fax: 01865 231870

☐ ***DNA analysis on blood sample only**
*If ophthalmoplegia is present an AChR ε mutation screen will be performed.
if no ophthalmoplegia *RAPSN* p.N88K and *DOK7* mutation screens will be undertaken.
If these are negative we recommend a clinic appointment to allow us to assess and direct further genetic testing**.**
If you approve of a clinic review if initial testing is negative please tick this box ☐

If specific mutational analysis is required, please send an email to David Beeson, clearly stating the required tests at: ***dbeeson@hammer.imm.ox.ac.uk***

Send DNA (10 ml EDTA or DNA extracted from EDTA blood) with pre-referral form to:

Duty Scientist, DNA Laboratory, Oxford Medical Genetics Laboratories
The Churchill Hospital, Headington, Oxford, OX3 7LJ.
http://www.ouh.nhs.uk/geneticslab

Please tick this box if the DNA sample has already been sent ☐

If you wish to **discuss the clinical case** please contact Dr Jackie Palace at:
jacqueline.palace@clinical-neurology.oxford.ac.uk

Patient name _____ Date of Birth _____ Sex M / F
First Surname or Family name

Address _____ Hospital ref. _____

_____ GP _____

_____ _____

Patient Post Code **(essential)**

_____

Patient Contact number:

_____

_____

NCG patient ID

_____

Genetic ID _____

DNA ID _____

Hosp. ID _____

Please leave blank for admin

Referring clinician _____

Address for correspondence _____

_____

_____

Date of referral: _____

Specific reason for referral: _____

I confirm that the patient, or parent/guardian if the patient is under age, has given permission for their DNA, and where relevant for that of family members, to be analysed for gene mutations that might be the cause of their myasthenic disorder.

To be signed by the referring clinician _____ _____
(please print name)

Pre-referral details (MUST BE PROVIDED IN ALL CASES)

Age at onset _____

		Delete
Predominant pattern of weakness :	Ptosis	Yes / No
	Extraocular muscles	Yes / No
	Feeding difficulties at birth	Yes / No
	Respiratory muscles	Yes / No
	Limbs	Yes / No
	Arthrogryposis	Yes / No

Other affected family members? _____

Results of investigations available to date:

Anti-AChR antibody titre: _____

Muscle biopsy: _____

_____

Tensilon or neostigmine test: _____

EMG: _____

Other: _____