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 ______________________________

Referring clinician ______________________________________________

Address for correspondence ______________________________________________

____________________________  ______________________________________

Patient Contact number: ______________________________  ______________________________

Patient Post Code (essential)  NCG patient ID

____________________________  ________________

Genetic ID ______________________________  

DNA ID ______________________________  

Hosp. ID ______________________________  

Please leave blank for admin

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 ______________________________

Predominant pattern of weakness:

Ptosis                   Yes / No
Extraocular muscles     Yes / No
Feeding difficulties at birth Yes / No
Respiratory muscles     Yes / No
Limb(s)                 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: _____________________________________________________________________________________