Diagnostic and Advisory Service for Rare Neuromuscular Diseases

# Oxford - Referral Centre for Congenital Myasthenic Syndromes (CMS)

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

This is currently a free service to patients living in England and 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 around 33 genes associated with comprising more than 500 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. This is performed in the immunology lab at the Churchill Hospital directly, or when we see the patient in the clinic (if felt appropriate)
* pre-natal screening

# Pre-Referral Form

**Please note: We cannot accept samples unless this is completed.**

**Two levels of service are offered. Please tick the box to indicate which you require below:**

**(Preferred)** Clinical assessment, specialised investigations and DNA analysis on blood sample.

**Return FORM ONLY to:** CMS Administrator, Department of Clinical Neurology, Level 3 West Wing, John Radcliffe Hospital, Headington, Oxford, OX3 9DU Telephone: 01865 231915

\*DNA analysis on blood sample only

If the main panel is negative, and you feel the patient has features of CMS 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 further specific analysis of variants of unknown significance (VUS) is required, please send an email clearly stating the required tests and reasons to** [**Yin.Dong@ouh.nhs.uk**](mailto:Yin.Dong@ouh.nhs.uk)

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

Mike Oldridge (DPhil), Oxford Regional Genetics Laboratories, Oxford University Hospitals NHS Foundation Trust, The Churchill Hospital, Oxford OX3, 7LE

**More information available at** [**www.ouh.nhs.uk/geneticslab**](http://www.ouh.nhs.uk/geneticslab)

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

Healthcare Professionals who wish to **discuss the clinical case,** please contact [**orh-tr.cmsgenetics@nhs.net**](mailto:orh-tr.cmsgenetics@nhs.net)

| **Patient Details** |  |
| --- | --- |
| First name |  |
| Surname or Family name |  |
| Date of birth |  |
| Sex | M / F |
| Address |  |
| Post code **(must be provided)** |  |
| Patient contact number |  |
| Hospital reference |  |
| GP |  |

| **Clinical Team Details** |  |
| --- | --- |
| Referring clinician |  |
| Address for correspondence |  |
| Date of referral |  |
| Specific reason for referral |  |

|  |  |
| --- | --- |
| **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  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 and whether RNS or SFEMG |  |
| Other |  |

**To be signed by the referring clinician**

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.

Signed \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Print name \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

| **FOR OFFICE USE ONLY** |  |
| --- | --- |
| NCG Patient ID | DNA ID |
| Genetic ID | Hospital ID |