MRC Centre for Neuromuscular Diseases

NHS England
Highly Specialised Services

NHS

# Diagnostic and Advisory Service for Rare Neuromuscular Diseases
# Institute of Neurology - Referral Centre for Ion Channel Disorders of Skeletal Muscle

## Pre - referral Form

This is a free service to patients living in **England** and **Scotland** . We are able to accept DNA samples from outside these areas, but charges will have to be made (please enquire).

The results and advice we are able to give you will be generated using a combined approach incorporating:

- Clinical information
- Electrophysiological assessment
- Analysis of DNA from a blood sample

These are needed to offer an informed opinion because of the heterogeneity within this group of disorders.

**Two levels of service are offered. Please indicate which you require below and then fill in ALL of the form on the reverse side.**

☐ **1. Letter and/or telephone advice plus DNA analysis**

☐ **2. Full clinical assessment, Electrophysiological assessment plus DNA analysis**

**Return form to**:
NCG Manager
National Hospital for Neurology & Neurosurgery
Box 102
Queen Square
London WC1N 3BG

**DNA Samples to**:
Neurogenetics
Laboratory
Room 630
Institute of Neurology
Queen Square
WC1N 3BG

**General Enquiries to**:
NCG Secretary
0203 448 8155
NCG Manager
0203 448 8030

If you wish to discuss the case before deciding which level of service you prefer please contact: Prof. Michael Hanna or Dr. Emma Matthews via the NCG channel email: musclechannelNCG@uclh.nhs.uk.

MRC | Centre for Neuromuscular Diseases

The National Hospital For Neurology And Neurosurgery

# Myotonia/Paramyotonia Congenita Proforma

| **Name:** | **Date of Birth:** | **Gender:** MALE / FEMALE |
|---|---|---|
| **Address** (postcode essential): | | **Hospital No:** |
| | | **Consultant**: |
| | | **Referring Hospital:** |

**Inheritance**: RECESSIVE ☐ DOMINANT ☐ (please attach FAMILY TREE if possible)

## SYMPTOMS

**Age of onset:**

**Distribution of myotonia:** ARMS ☐ LEGS ☐ EYES ☐ FACE ☐ JAW ☐

**Associated weakness:** TRANSIENT ☐ PROGRESSIVE ☐

**Myotonia characteristics:**

- WARM UP ☐
- MARKED WORSENING WITH COLD ☐
- WORSENING WITH EXERTION ☐
- K+ EXACERBATION ☐

**Adverse anaesthetic events & problems in pregnancy**

**Treatment:**

## EXAMINATION

**Dysmorphism:**

**Limbs:** HYPERTROPHY ☐ WEAKNESS ☐ ATROPHY ☐ NORMAL ☐

**Myotonia**: LIDLAG ☐ GRIP ☐ EYELIDS ☐ PERCUSSION ☐ JAW ☐

**Reflexes** : NORMAL ☐ REDUCED ☐ BRISK ☐ ABSENT ☐

**Gait:**

**Other:**

## INVESTIGATIONS

| **Blood tests:** CK | TFTs | DM1 analysis | DM2 analysis |
|---|---|---|---|

**ECG:** (Any evidence of long QT?)

**EMG/NCS:**

**MUSCLE BIOPSY**

**NCG ID:** **DNA No:** **GENETIC ID:**

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# Periodic Paralysis Proforma

| **Name:** | **Date of Birth:** | **Gender:** MALE / FEMALE |
|---|---|---|
| **Address** (postcode essential): | | **Hospital No:**<br>**Consultant**:<br>**Referring Hospital:** |

**Inheritance**: DOMINANT ☐ RECESSIVE ☐ (please attach FAMILY TREE if possible)

## SYMPTOMS

**Age of onset:**

**Distribution of weakness:** ARMS ☐ LEGS ☐
BULBAR ☐ FACE ☐
RESPIRATORY ☐

**Potassium level during attack:**

**Frequency of attacks:**

**Associated Myotonia:** YES ☐ NO ☐

**Adverse anaesthetic events & problems in pregnancy:**

**Treatment:**

**Precipitant of attacks:**

REST AFTER EXERCISE ☐
STRESS ☐
CARBOHYDRATE MEAL ☐
ALCOHOL ☐ ----------
OTHER
WARM UP? ☐
WORSENING WITH EXERTION ☐
MARKED WORSENING WITH COLD ☐

## EXAMINATION

**Dysmorphism:**

**Limbs:** HYPERTROPHY ☐ ATROPHY ☐
NORMAL ☐
**WEAKNESS** ----------

**Reflexes** : NORMAL ☐ REDUCED ☐
BRISK ☐ ABSENT ☐

**Myotonia**: LIDLAG ☐ GRIP ☐
EYELIDS ☐ PERCUSSION ☐
JAW ☐

**Gait**:

**Other:**

## INVESTIGATIONS

| **Blood tests:** CK | TFTs | DM1 analysis | DM2 analysis |
|---|---|---|---|

**ECG:**

**EMG/NCS:**

**MUSCLE BIOPSY**

**NCG ID:** **DNA No:** **GENETIC ID:**