Highly Specialised Services - Muscle Channelopathy Service
National Hospital for Neurology and Neurosurgery
Box 102, Queen Square, London, WC1N 3BG
Email: uclh.enquiry.musclechannelservice@nhs.net

QUEEN SQUARE CNMD
Centre for Neuromuscular Diseases

Diagnostic and Advisory Service for Muscle Channelopathies

Referral Form

Referrals to the Diagnostic service:

We are happy to accept any adult referrals for possible muscle channelopathies in our Channelopathy Clinic at the National Hospital for Neurology and Neurosurgery. This just requires an e-referral from a GP or letter from a hospital doctor and is free for patients in England and Scotland. It is also available to patients in other areas with prior authorisation (please enclose the funding authorisation letter with the referral) . Children under 16 years with possible muscle channelopathies can be seen in our Paediatric Channelopathy Clinic at Great Ormond Street Hospital with a GP or hospital doctor referral.

Referral criteria for Genetic testing only:

We can accept referrals for the genetic testing of the skeletal muscle channelopathy panel (R76.1) from Neurologists or Clinical Geneticists (as per the National Genomic Test Directory eligibility criteria: https://www.england.nhs.uk/publication/national-genomic-test-directories/).

All requests require a completed referral form before the sample can be tested and must meet the following criteria:

1. **Adults**:
    - Patient consent for testing
    - Signs or symptoms of a muscle channelopathy
      Or
    - Family history of a muscle channelopathy
2. **Children** (under 16 years)
    - Signs or symptoms of a muscle channelopathy
    - Parental consent for testing

   (Please note we do not test asymptomatic children except in cases of a family history of Andersen Tawil syndrome)

Contact details:

Please send all genetic samples to:
Rare & Inherited Disease Genomic Laboratory
North Thames GLH
Level 5 Barclay House
37 Queen Square
London WC1N 3BH
Email: ucl-tr.NHNNgenetics@nhs.net
General Genetic Tel Enquiries: 0207 829 8870

All referral forms can be sent to:
Email: uclh.enquiry.musclechannelservice@nhs.net

For any clinical advice or queries or to discuss referrals please contact:
Dr Dipa Jayaseelan: dipa.jayaseelan@nhs.net
Professor Mike Hanna: michael.hanna@nhs.net

Clinical Enquiries: 020 3448 8155

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Diagnostic and Genetic Referral Form for Muscle Channelopathies

Please fill out this form if you would like patients to be seen for clinical analysis or muscle channel genetic analysis.

Patient and Requester Details

NHS No:		**Sex:**		**Date of Birth:**	
Surname:		**Address (including postcode):**			
Forename:					
Ethnicity:		**Clinician:**			
Hospital:		**Hospital No:**			
Email:		**Reporting address:**			

Assessment Information

Clinical Assessment Required?	☐
Genetic Testing Required?	☐

Possible Clinical Diagnosis

Periodic Paralysis	☐
Myotonia	☐

Clinical Details

Age at onset:	
Myotonia?:	Yes: ☐ No: ☐
Warm-up	☐
Cold sensitive	☐
Worse with exercise	☐
Distribution of myotonia?	Yes: ☐ No: ☐
Face/eyes	☐
Arms	☐
Legs	☐
Weakness?	Yes: ☐ No: ☐
Episodic	☐
Progressive	☐
Distribution of weakness	
Precipitants?	
Anaesthetics problems	Yes: ☐ No: ☐ Details:
Previous treatments	
Family history:	Yes: ☐ No: ☐
Consanguinity:	☐

Examination

Muscle weakness:	Yes: ☐	No: ☐
Clinical myotonia:	Yes: ☐	No: ☐
Hypertrophy:	Yes: ☐	No: ☐
Hypotonia	Yes: ☐	No: ☐
Facial dysmorphism	Yes: ☐	No: ☐
Other signs (e.g. contractures, pain):		

Results of Investigations

CK:	**K+ during attack:**	
DM1 testing: ☐	DM2 testing: ☐	
ECG:	Normal ☐	Long QT ☐
EMG:	Myotonia ☐	Myopathy ☐
Short exercise test:		
Long exercise test:		
Muscle biopsy:		

Relevant correspondence and other information (Include additional clinical features and family tree)

Website: www.channelopathy.nhs.uk
Admin Office: 020 3448 8155
Clinical queries: Dr Dipa Jayaseelan (dipa.jayaseelan@nhs.net)

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