Diagnostic and Advisory Service for Rare Neuromuscular Diseases



##### **National Service Referral Form for**

##### **Congenital Muscular Dystrophies and Myopathies**

The Highly specialised Service (HSS) for congenital muscular dystrophies and myopathies is a nationally funded service based at the Dubowitz Neuromuscular Centre in Great Ormond Street Hospital in London and is available free of charge to patients living in England and Scotland. We are able to accept referrals from outside England and Scotland, but charges will have to be made (please enquire).

**FOUR LEVELS OF SERVICE ARE OFFERED (*tick as appropriate)***

* Clinical assessment
* Muscle biopsy (stained slides or frozen muscle) analysis
* Histological, histochemical, immunohistochemical and electron microscopy
* Collagen 6 flow cytometry analysis on skin fibroblast cultures
* DNA analysis (see 2nd page for details)

**THE REFERRAL PACK SHOULD INCLUDE:**

* **Detailed clinic letters (mandatory)**
* **Muscle biopsy report (mandatory)**
* MUSCLE MRIImages on CD/via GOSH Image Exchange Portal (desirable)
* BRAIN MRI Images on CD/via GOSH Image Exchange Portal (desirable)
* CLINICAL PHOTOGRAPHS (if available/indicated)

**Referrals for clinical assessments** should be sent to: Prof F Muntoni or Dr A Sarkozy at Level 9, Nurses Home, Great Ormond Street Hospital, London WC1N 3JH. If you wish to discuss the request, please contact Dr. A Sarkozy [anna.sarkozy@gosh.nhs.uk](mailto:anna.sarkozy@gosh.nhs.uk), [anna.sarkozy@nhs.net](mailto:anna.sarkozy@nhs.net); Prof. F Muntoni ([francesco.muntoni@gosh.nhs.uk](mailto:francesco.muntoni@gosh.nhs.uk)) or the HSS coordinator Mrs Gombosed [gundari.gombosed@nhs.net](mailto:gundari.gombosed@nhs.net) at Tel: 020 7405 9200 ext 5849

**Referrals for muscle biopsy/slides analysis and Collagen 6 FACS analysis** should be sent to: Dubowitz Neuromuscular Centre (DNC), 1st Floor Department of Neuropathology, Institute of Neurology, Queen Square House, Queen Square, London WC1N 3BG – Tel: 020 3448 4235 and/or email to juliane.mueller@gosh.nhs.uk or francesco.catapano@gosh.nhs.uk. If you wish to discuss the pathological features, please contact: Dr. R Phadke [rahul.phadke@nhs.net](mailto:rahul.phadke@nhs.net) Tel: 020 3448 4393. For general enquiries email: [gos-tr.dnc.musclepathology@nhs.net](mailto:gos-tr.dnc.musclepathology@nhs.net).

**~ Please, telephone the laboratory prior to dispatching the samples ~**

**~Site of the biopsy must be clearly indicated on the form~**

**Referrals DNA analysis** **(@Oct2020)** should be sent directly to: DNA Laboratory, Viapath Genetic Centre, 5th Floor Tower Wing, Guy's Hospital, St Thomas Street, London SE1 9RT – Tel: 020 7188 2582/1714. If you wish to discuss a request, please contact Viapath [gst-tr.ViapathGeneticsAdmin@nhs.net](mailto:gst-tr.ViapathGeneticsAdmin@nhs.net) or Dr. A Sarkozy [anna.sarkozy@gosh.nhs.uk](mailto:anna.sarkozy@gosh.nhs.uk), [anna.sarkozy@nhs.net](mailto:anna.sarkozy@nhs.net) Tel: 020 7405 9200 ext 5849.

**Referrals must include the following information:**

1. **Please select the appropriate panel according to clinical indication:**

* R79 Congenital Muscular dystrophy (PanelApp number 207)

https://panelapp.genomicsengland.co.uk/panels/207/

* R81 Congenital Myopathy (PanelApp number 225)

<https://panelapp.genomicsengland.co.uk/panels/225/>

*See* [*https://panelapp.genomicsengland.co.uk/panels/*](https://panelapp.genomicsengland.co.uk/panels/) *for genes included in the analysis.*

*While analysis will include all “green genes” listed in PanelApp, knowledge of the most likely causative gene/s would aid genetic analysis and variant interpretation. If possible, please select from or add in the list below, as appropriate:*

*❒ COL6 and COL12 genes*

*❒ LAMA2 gene*

*❒ Genes responsible for Alpha-dystroglycanopathies*

*❒ Genes responsible for Core myopathies*

*❒ Genes responsible for Centronuclear myopathies*

*❒ Genes responsible for Nemaline myopathies*

*❒ ...................................................................................................................................*

1. **Testing is activated in accordance with eligibility criteria indicated in the NHS England National Genomic Test directory for rare and inherited disease** [**https://www.england.nhs.uk/publication/national-genomic-test-directories**](https://www.england.nhs.uk/publication/national-genomic-test-directories)**). Please select as appropriate for your patient:**

* Clinical features of congenital myopathy
* histopathological features of congenital myopathy
* muscle MRI features of congenital myopathy
* other features of congenital myopathy (please add details at page 3-5)
* Clinical features of Congenital muscular dystrophy
* brain or muscle MRI features of Congenital muscular dystrophy
* histopathological features of congenital muscular dystrophy
* other features of congenital muscular dystrophy (please add details at page 3-5)

1. **DNA referrals must include a completed referral form, detailed clinic letters and muscle biopsy report (if available)**

*The results and specialist advice we are able to give you will be generated using a combined approach incorporating clinical information, specialised analysis of muscle biopsy, MRI review in selected cases and genetic analyses. The combination of these approaches allows offering an informed and specialist opinion because of the heterogeneity within this group of disorders. Thus, we would recommend providing all supporting information at time of referral, to speed up analytical and post-test interpretation process. Thank you*

**DNC Lab use ONLY**

**BIOPSY No:**

# DATE & TIME SAMPLE RECEIVED:

# 

**PATIENT INFORMATION**

Hosp. Nº: NHS Nº: Male/Female: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Forename: Surname: D.O.B: \_\_\_\_\_\_\_\_\_\_\_\_\_\_

Address: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Referring clinician: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Patient Post Code **(essential)**

Address for correspondence:

Specific reason for referral:

**PRELIMINARY CLINICAL DIAGNOSIS AT REFERRAL**:

❒ **Is result required urgently?** If so, please state reason and time frame\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**DETAILS OF SPECIMEN:** (see accompanying document on how to collect & send clinical samples)

# ❒ blood ❒DNA ❒muscle biopsy OCT Block ❒skin biopsy ❒cell culture ❒ slides ❒CVS

**DATE OF BIOPSY (Essential)**:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**SITE OF BIOPSY (Essential):** \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**TYPE OF BIOPSY**: ❒open ❒needle ❒fresh ❒frozen (OCT block preferred)

**PATHOLOGICAL FINDINGS** (please enclose report) **\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**Has the sample been consented for storage?** ❒Yes muscle**/** fibroblasts**/** blood**/** DNA ❒No

**Has the sample been consented for research?** ❒Yes muscle**/** fibroblasts**/** blood**/** DNA ❒No

# CLINICAL INFORMATION: (tick where appropriate)

Onset age: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Symptoms at onset\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ decreased foetal movements ❒ polyhydramnios ❒ weeks of gestation \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ hypotonia ❒ congenital hip dislocation ❒ joint laxity at birth

❒ congenital contractures (describe):\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ delayed motor milestones

❒ AGE AT SITTING: \_\_\_\_\_ AGE AT WALKING: \_\_\_\_\_\_

CURRENT FUNCTIONAL LEVEL: ❒Ambulant and able to climb stairs

❒Unable to climb stairs

❒Uses wheelchair full time

MAXIMAL MOBILITY ACHIEVED (if different):\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

AGE AT LOSS OF AMBULATION: \_\_\_\_\_\_\_\_\_\_

❒MUSCLE WEAKNESS: ❒PROXIMAL UPPER LIMBS ❒DISTAL UPPER LIMBS

❒PROXIMAL LOWER LIMBS ❒DISTAL LOWER LIMBS ❒AXIAL ❒BULBAR ❒FACIAL

❒PTOSIS ❒OPHTALMOPLEGIA

(Describe pattern of weakness & age at onset): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒FEEDING DIFFICULTIES ❒FAILURE TO THRIVE ❒GASTROSTOMY

❒SCOLIOSIS ❒ SPINAL RIGIDITY ❒ SKIN FEATURES (describe)\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ JOINT LAXITY ❒POSTNATAL CONTRACTURES, (describe): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒CARDIOMYOPATHY ❒ CARDIAC CONDUCTION DEFECT

❒RESPIRATORY (FVC value, recurrent chest infections, FVC value, ventilatory support): \_\_\_\_\_\_\_\_\_\_\_\_\_

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ MUSCLE WASTING (describe):\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ MUSCLE HYPERTROPHY (describe): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒ MUSCLE MRI (describe/enclose report): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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❒STRUCTURAL EYE INVOLVEMENT ❒ Cataracts ❒ MALIGNANT HYPERTHERMIA REACTION

❒ LEARNING DIFFICULTY ❒EPILEPSY ❒ MICROCEPHALY

❒ BRAIN MRI (describe/enclose report):\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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❒ EMG /NCV (describe/enclose report, specify if SFEMG done): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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❒ ADDITIONAL FINDINGS:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒CK LEVELS: \_\_\_\_\_\_\_\_\_\_\_\_\_AT AGE:\_\_\_\_\_\_\_\_\_\_\_\_\_ Normal upper limits in your lab:\_\_\_\_\_\_\_\_\_\_\_\_\_\_

❒PREVIOUS GENETIC TESTS in particular please indicate if DNA was already submitted for NGS analysis to NHS laboratory: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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❒WAS THE PATIENT ENROLLED IN RESEARCH STUDIES? (if yes, please detail): \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**FAMILY HISTORY**:

❒ CONSANGUINITY

❒ OTHER AFFECTED FAMILY MEMBERS:

❒ NAME AND RELATIONSHIP OF OTHER AFFECTED MEMBERS (include clinical and biopsy details if possible)

❒ ETHNICITY

❒ PEDIGREE:

❒ Any additional information: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Signed: Date: