

Diagnostic and Advisory Service for Rare Neuromuscular Diseases

Newcastle upon Tyne - Referral Centre for Limb Girdle Muscular Dystrophy
Funded through NHS England Highly Specialised Services for Rare Neuromuscular Diseases and the Specialised Health Services
Commission for Wales

LGMD service details

The nationally funded service for Limb Girdle Muscular Dystrophy (LGMD) is based in Newcastle upon Tyne and is available **free of charge** to patients living in England, Scotland and Wales. We are also able to accept biopsies and DNA samples from outside these regions, as well as private patient referrals - please enquire regarding specific costs for different aspects of our service.

- Protein analysis of frozen muscle samples supports genetic analysis of a DNA sample and is also useful in characterising unclassified variants (UCVs). We are able to accept biopsy samples from diagnosed DMD/BMD patients for dystrophin quantification.
- Demonstration of protein levels and localisation using immunohistochemistry (IHC) and western blotting (WB) techniques can reveal primary genetic defects and can also show a pattern or profile of secondary alterations that contribute to a suggested diagnosis. A panel of antibodies is used to analyse proteins - please enquire for details of current antibodies used.
- All muscle biopsies and molecular results are discussed at a weekly Advisory MDT and an advisory letter will follow. You are welcome to join the MDT to discuss your referral when results are available.
- Any clinical referrals should be sent FAO Dr Chiara Marini-Bettolo. A clinical appointment in our new patient clinic will be provided.
- We accept referral of paediatric and adult patients with suspected diagnosis of LGMD and distal myopathies or a confirmed diagnosis where a second opinion or access to research is requested.

What to do when sending a biopsy

It is essential that the Pre-referral form is completed for all biopsy referrals

- Samples referred without the current version of the Pre-referral form may have analysis **delayed** as we may require additional information before processing.
- It would be helpful if you could provide MRI images and/or results of any previous relevant muscle biopsy analysis or DNA testing to aid interpretation.
- If a biopsy referral is deemed to be inappropriate, the referring centre will be informed to avoid wasting any material.

Please note that tissue samples from patients known or suspected of being infected with Hazard Group 3 or 4 organisms (such as TB, HIV or Hepatitis) **must not be sent** for protein analysis.

Sample requirements

Analysis is performed on **unfixed frozen muscle** - ideally samples should measure approximately 3mm x 3mm x 3mm (25 mg) - the minimum required for immunoanalysis. All samples must be clearly labelled (with both the patient's name and date of birth, which must also be written on the accompanying paperwork).

Dispatch instructions for muscle biopsies

Please read this section carefully – muscle biopsies are extremely precious specimens and failure to follow these instructions may result in samples being delayed, lost or damaged. Please note; samples cannot be received on Fridays.

1. Please **telephone** the laboratory to confirm when the sample will arrive.
2. Pack the sample in a thick walled polystyrene container, fill with sufficient dry ice to last 48 hours (at least 5kg) and pack into a cardboard box marked with appropriate hazard and shipping labels.
3. Arrange a courier for **door-to-door delivery** of the sample and sample identification documentation to:

LGMD Referral Centre, Muscle Immunoanalysis, Lower Ground Floor (Room 2.026), Dental Hospital, Richardson Road, Newcastle upon Tyne, NE2 4AZ

Service contact details

General enquiries & referrals nuth.hss.lgmd.referrals@nhs.net

Ms Susan Robinson	Administrator	susan.robinson27@nhs.net	0191 282 0841
Mr Matt Henderson	Lead Biomedical Scientist	m.henderson7@nhs.net	0191 282 0847
Mrs Yolande Parkhurst	Biomedical Scientist	yolande.parkhurst@nhs.net	0191 282 0842

NSCT National Referral Centre for Limb Girdle Muscular Dystrophy ■ Head of Service: Dr Chiara Marini-Bettolo

A Laboratory form containing data is NOT a controlled document

Valid on day of print only:

The Proforma is a Controlled document. Document details i.e. Update responsibility, Ultimate approver, Active date and Review date are held in Q-Pulse. If you recognise an inaccuracy or can suggest an improvement, please raise a Change Request on Q-Pulse

Muscle Immunoanalysis Unit

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National Referral Centre for Limb Girdle Muscular Dystrophy

Immunoanalysis Pre-Referral Form

Each request received by the laboratory is considered an agreement between the laboratory and the sender. Consent is implied for the processing and storage of any samples received

- Please complete the form electronically and submit (along with copies of clinical letters etc.) via email for review
- All essential fields (denoted by *) **must** be completed. Failure to do so **may** result in a delay to analysis being performed

LGMD service options*

Please indicate which level of service you require

- Muscle biopsy analysis ☐
- Review of stained slides ☐

Is a result required **URGENTLY**? ☐

Reason for urgency

Office Use only

Date FORM received	
Date BIOPSY received	
Biopsy ID	H
GC Identifier	GC

Patient Details

SURNAME*

Forename*

DOB*

Gender* Choose option

NHS No.*

Hospital No.

Address*

Postcode*

Referring clinician details*

Name

Designation

Address for correspondence

Muscle sample details

Site of muscle biopsy

Date biopsy collected

Mandatory clinical information*

Analysis could be delayed until all of this information is provided

Working diagnosis

CK level

at age yrs.

Age at onset

yrs.

Disease course: Choose option

Other relatives affected Choose option

Inheritance Choose option

Consanguinity Choose option

Functional ability

Ambulant ☐

Able to walk on toes ☐

Able to walk on heels ☐

Able to climb stairs ☐

Weakness distribution

Proximal upper limbs ☐

Proximal lower limbs ☐

Distal upper limbs ☐

Distal lower limbs ☐

Other

Clinical information checklist*

Please confirm which of the following information has been submitted with this referral

Clinical letters ☐

Muscle MRI ☐

Image access via PACS portal (to The Newcastle upon Tyne NHS Foundation Trust)

Muscle biopsy report ☐

Genetics report(s) ☐

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Other clinical information

Delayed motor milestones ☐

Cognitive dysfunction / intellectual disability ☐ If yes: Age at onset yrs.

EMG results Normal ☐ Please attach reports if available and include details in Additional comments (below)

Nerve conduction velocities ☐ Detection EMG ☐ Repetitive nerve stimulation ☐ Single fibre EMG ☐

Atrophic muscles

Proximal upper limbs ☐ Proximal lower limbs ☐ Distal upper limbs ☐ Distal lower limbs ☐

Hypertrophic muscles

Proximal upper limbs ☐ Proximal lower limbs ☐ Distal upper limbs ☐ Distal lower limbs ☐

Asymmetry ☐

Skeletal deformities

Contractures ☐ Scoliosis ☐ Scapular winging ☐ Spinal rigidity ☐
If contractures: Proximal ☐ Distal ☐

Ocular signs

Ptosis ☐ Ophthalmoparesis ☐ Cataracts ☐

Associated features

Speech problems ☐ Swallowing problems ☐ Hearing loss ☐ Neuropathy ☐
Muscle pain ☐ Cramps ☐ Rippling muscles ☐ Skin abnormalities ☐
Bone abnormalities ☐

Cardiac abnormalities

Cardiomyopathy ☐ Arrhythmia ☐ Conduction defect ☐

Respiratory impairment

Current / past medication

Brain CT scan / MRI ☐ Result

Additional comments



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