Repertoire of Tests

1.BIOPSY SERVICE

Repertoire of Tests

The tests that have been carried out will be specified in the report.
If slides with good staining quality are supplied some techniques may not be repeated

1.1 Muscle Biopsy

Histology and Histochemistry

Routine tests:
Haematoxylin & Eosin (H&E)
Gomori modified Trichrome (TRI)
Reduced nicotinamide adenine dinucleotide tetrazolium reductase (NADH-TR)
Succinate dehydrogenase (SDH)
cytochrome oxydase (COX)
Combined COX/SDH
PAS Schiff's reagent (PAS)
Oil Red O (ORO)

The following tests will be performed as appropriate to the clinical phenotype and/or after initial analyses.

Additional tests:
Acid phosphatase
Phosphofructokinase (PFK)
Myophosphorylase
Myoadenilate deaminase (AMP)
Menadione-linked α-glycerophosphate dehydrogenase (MGD):

Immunohistochemistry

Routine tests:
Dystrophin (dys- 2 and -3)
β-spectrin
Utrophin
Neuronal nitric oxide synthase (nNOS)
Neonatal myosin heavy chain
Fast myosin heavy chain
Slow myosin heavy chain
β-dystroglycan
α-dystroglycan (clone IIH6)
Sarcoglycans (α,β,)
Collagen VI (MAB 1944 and MAB 3303) and double labelling with perlecan if appropriate

Laminin α2 (80kDa, MAB1922, 300 kDa, Alexis 4H8)
Laminin α5
Laminin β1
Laminin γ1
Major histocompatibility complex type 1 (MHC-1)

Additional tests (when appropriate):
Laminin β2 MDC1A
NCAM
Dysferlin (Hamlet, Hamlet-2)
Emerin
Lamin A, Lamin A/C, Lamin B1 and Lamin B2
Laminin-α2 (NCL-Mer antibody)
Myotilin,
Caveolin3
Desmin
Cardiac actin,
Nebulin
Actin
α-actinins
Serca1 and secra 2

The diagnostic value of immmunoblotting is currently being assessed for

α-dystroglycan (IIH6)
β-dystroglycan
Laminin-α2 (MAB1922)
RYR1

Electron microscopy:
Ultrastructural analysis of muscle. This is an important technique in the differential diagnosis of congenital myopathies. We will accept the resin block, grids or photographs.

Assessment of stained slides for second opinion

Slides can be referred either on their own, or prior to referring the muscle biopsy. Please send stained sides with a copy of the pathology report if available.

1.2 Skin Biopsy

Immunohistochemistry on frozen skin OCT block
Laminin-α2 (80kDa MAB1922, 300 kDa Alexis 4H8)
Laminin -α5
Laminin -β1
Laminin -γ1
Laminin ß2

Additional:
Laminin -α2 (NCL-Mer antibody)
Emerin

1.3 Skin Fibroblast
Immunohistochemistry
Collagen VI, (antibodies: clone MAB 1944 and MAB 3303

1.4 C.V.S.

Immunohistochemistry
Collagen VI (MAB1944, MAB 3303)

Laminin-α2 (80kDa MAB1922, 300 kDa Alexis 4H8)
Laminin -α5
Laminin -β1
Laminin -β2
Laminin -γ1

2. DNA ANALYSIS

Repertoire of Tests

2.1 Sequencing for Mutations (genes)
LAMA2 (MDC1A)
SEPN1 (RSMD1)
POMT1/POMT2
FKRP
POMGnT1
LARGE
Fukutin
COL6A1 & COL6A2 & COL6A3
RYR1
ACTA1
MTM1sequencing + MLPA
BIN1
DNM2
TPM2
TPM3
CFL2
NEB exon 55 deletion + linkage

2.2 Haplotype analysis at the corresponding loci for the following forms of congenital muscular dystrophy:
UCMD: only for families with known recessive inheritance since dominant and *de novo* mutations are common -
MDC1A