

Final

Great Ormond Street Hospital for Children NHS Trust

NCG National Referral Centre for Congenital Muscular Dystrophies and Myopathies

Head of Service: Prof. F. Muntoni

DNC biopsy service repertoire of tests

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 (PAS)

PAS after diastase (DPAS)

Oil Red O (ORO)

Acid phosphatase

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

Additional tests:

Phosphofructokinase (PFK)

Myophosphorylase

Myoadenilate deaminase (AMP)

MGD (menadione-linked α -glycerophosphate dehydrogenase without substrate):

Immunohistochemistry

Routine tests:

Dystrophin (dys-1,2 and -3)

β -spectrin

Utrophin

Neuronal nitric oxide synthase (nNOS)

Foetal myosin

Developmental Myosin

Fast myosin heavy chain

Slow myosin heavy chain

β -dystroglycan

α -dystroglycan (clone IIH6)

Sarcoglycans (α , β , γ , δ)

Collagen VI (MAB 1944 and MAB 3303) and double labelling with perlecan

Laminin α 2 (80kDa, MAB1922, 300 kDa, Alexis 4H8)

Laminin α 5

Laminin β 1

Laminin γ 1

Major histocompatibility complex type 1 (MHC-1)

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NCAM

Caveolin3

Dysferlin (Hamlet1)

Desmin

P62

Additional tests (when appropriate):

Laminin β 2

Emerin

Lamin A, Lamin A/C, Lamin B1 and Lamin B2

Myotilin,

alpha B-Crystallin

Filamin C

FHL-1

BAG3

Cardiac actin

Plectin

Phalloidin

Nebulin

Actin

α -actinins

Serca1 and Serca 2

CD3

CD4

CD8

CD20

C5B-9 (MAC)

Nesprin (1 and 2)

Lamp-2

Calpain3

Collagen III

Collagen IV

Collagen V

Electron microscopy (EM):

Ultra-structural analysis of muscle. This is an important technique in the differential diagnosis of congenital myopathies. We will accept the resin blocks, grids, EM prints or EM images on a CD.

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Assessment of stained slides for second opinion

Stained slides can be referred either on their own, or prior to referring the muscle biopsy. If available please send stained slides if a frozen block is referred, with a copy of the pathology report