

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

Sarcoglycans ($\alpha, \beta, \gamma, \delta$)

Laminin $\alpha 2$ (80kDa, 300 kDa), $\alpha 5, \beta 1, \gamma 1$

Major histocompatibility complex type I (HLA-ABC)

NCAM

Caveolin3

Dysferlin (Hamlet1)

Desmin

P62

Additional tests (when appropriate):

α -dystroglycan (clone IIH6)

Collagen VI (chain specific A1, A2, A3) and double labelling with perlecan

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

Major histocompatibility complex type II (HLA-DP,DQ,DR)

CD3

CD4

CD8

CD20

CD31

CD68

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 or images sent electronically.

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