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The role of Immunohistochemistry for evaluation of Muscle Biopsies

Evaluation of a muscle biopsies is an important part of the diagnostic pathway of a neuromuscular disorder, even though next generation sequencing is now more widely used. Muscle biopsy can contribute to the selection of appropriate gene panels, to the assessment of the pathogenicity of gene variants and identify specific pathological features when next generation sequencing has produced negative results. Immunohistochemistry is an essential tool for the assessment of a muscle biopsy, but the data must always be correlated with the clinical, biochemical, electrophysiological, imaging, histological and histochemical results. The localisation of antibodies by immunohistochemistry can identify specific protein abnormalities resulting from a primary defect in a gene, for example the absence of a particular sarcolemmal protein can often identify the form of muscular dystrophy. Studies of secondary abnormalities also have a role, such as the over-expression of a protein in response to a primary defect in a gene. In acquired, non-inherited neuromuscular disorders immunohistochemistry can identify several informative abnormalities that aid diagnosis, direct appropriate therapy and aid differential diagnosis. Immunohistochemical studies of antibodies to myosin heavy chain isoforms are now recognised as an important tool for the assessment of fibre types in both inherited and acquired muscle disorders, for assessing maturity of fibres and for determining if a muscle sample is normal or abnormal.

Recent Publications:

1. Importance of immunohistochemical evaluation of developmentally regulated myosin heavy chains in human muscle biopsies. Sewry CA, Feng L, Chambers D, Matthews E, Phadke R.Neuromuscul Disord. 2021 May;31(5):371-384.DOI: 10.1016/j.nmd.2021.02.007.

2. Nemaline myopathies: a current view. Sewry CA, Laitila JM, Wallgren-Pettersson C.J Muscle Res Cell Motil. 2019 Jun;40(2):111-126. doi: 10.1007/s10974-019-09519-9. Epub 2019 Jun 21.

Biography

Caroline Sewry has worked in the field of muscle pathology for over 50 years. She is a clinical scientist who did her first degree in Zoology and PhD in electron microscopy of muscle. She has worked closely with Professor Victor Dubowitz and Professor Francesco Muntoni, two of the leading paediatricians in the neuromuscular field. In 1998 she established a muscle biopsy service at the RJAH Orthopaedic Hospital, Oswestry where she continues to work part time and also works part time at Salford Royal Hospital, Manchester. She is co-author of the 3rd, 4th and 5th editions of 'Muscle Biopsy A Practical Approach' and the chapter on muscle disorders in the 8th and 9th edition of Greenfield's Neuropathology. She is co-editor of 'Muscle Disease Pathology and Genetics 2nd edition, an author of over 300 peer reviewed papers, as well as contributor to chapters in several textbooks on muscle disease.

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