Muscle Biopsy Findings in Mitochondriopathies

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The mitochondrial myopathies are a heterogeneous group of neuromuscular disorders in which abnormalities in mitochondrial function may be associated with structural abnormalities in the mitochondria. During recent years the field of mitochondrial myopathies has been grown with recognition of a maternal pattern of inheritance in some syndromes and an autosomal dominant or recessive pattern in others. Mitochondrial myopathies may be overlooked on routine histological stains of muscle but may be suspected by the presence of 'ragged-red fibers' on the Gomori trichrome stain. Peripheral mitochondrial proliferation happen in the mitochondrial myopathies and it may be difficult to differentiate from those that occur to varying degrees in normal muscle. Careful examination of the three oxidative enzyme reactions (NADH-TR, SDH and COX) is required and combining the demonstration of SDH with COX on the same section can be helpful in identifying fibers devoid of COX, as they appear blue. Abnormal fibers may show excessive lipid and glycogen accumulation. The number of ragged-red or COX-negative fibers is variable, ranging from many to few, irrespective of the degree of clinical involvement.

Ragged-red fibers and fibers devoid of COX are not seen in all cases of mitochondrial myopathy and are more commonly associated with mutations in mitochondrial DNA rather than the nuclear genes. It can help the diagnosis of Kearns-Sayre syndrome, Progressive external ophthalmoplasia, Pearson syndrome, MERRF, MELAS as well as complex II deficiency, CoQ10 deficiency, COX deficiency. It must also be remembered that ragged-red fibers may occur as a secondary change in other cases, such as muscular dystrophies and inflammatory myopathies like Inclusion Body Miositis (IBM).

Keywords: Mitochondriopathy; Mitochondrial myopathy; Muscle biopsy; Ragged-red fiber; COX-negative fiber

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