

Is muscle biopsy still necessary for the diagnosis of mitochondrial disease?

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Mitochondria are essential organelles for human life and play a central role in diverse cellular processes such as energy production, reactive oxygen species, biosynthetic intermediates, autophagy, amino acid and fatty acid synthesis and degradation, heme synthesis, intracellular calcium homeostasis and apoptosis in physiological and pathological conditions⁽¹⁾.

Mitochondrial diseases are an inherited genetic group of disorders characterized by abnormal oxidative phosphorylation that results from mitochondrial deoxyribonucleic acid (mtDNA) or nuclear (n)DNA genes giving rise to a constellation of multisystemic manifestations and a wide spectrum of neurological manifestations (metabolic myopathy, seizures, ataxia, encephalopathy, movement disorders, peripheral neuropathy and psychiatric disturbances) from birth to senescence^(2,3).

Skeletal muscle is an ancestral source of accessing and studying mitochondrial dysfunction in primary mitochondrial diseases and in other conditions, such as neurodegenerative or inflammatory disorders and in physiological processes like aging. Classical morphological findings such as ragged-red fibers (RRF), blue fibers (BF) and cytochrome c oxidase negative (COX-) fibers are in the imaginary and biological references of various health professionals and remained as diagnostic markers until the identification of the first genetic alterations in mitochondrial DNA in 1988^(4,5).

Nowadays, the omics techniques (genomics, transcriptomics, proteomics, metabolomics and epigenomics), with advanced bioinformatics tools, have produced a revolution in mitochondrial functionalities and their relationship with cellular health and disease, providing novel knowledge for the diagnosis and therapeutics of mitochondrial diseases⁽⁶⁾.

In this edition of *Jornal Brasileiro de Patologia e Medicina Laboratorial* (JBPM), Kouyoumdjian *et al.* (2018)⁽⁷⁾ present a symbolic and needful study about the role of classical pathological skeletal muscle biopsy features according to Walker and Sleigh pathological criteria for mitochondrial disorders in the molecular era of mitochondrial studies. The paper shows that some morphological features, such as RRF, BF, COX- and COX staining with succinate dehydrogenase (COMBO+), are still useful and valid as ancillary diagnostic methods in the diagnostic odysseys of patients suffering from mitochondrial diseases and are paving the way for a more complete knowledge in mitochondrial medicine.

We wish you enjoy the reading!

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