

ULTRASTRUCTURAL CHANGES IN MITOCHONDRIAL MYOPATHY

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Introduction: The mitochondrial myopathy can be in relation to genetic alteration in mitochondrial or nuclear DNA. They are multisystemic dysfunctions that affect organs that depend on the mitochondrial energy, as central nervous system, heart and skeletal muscle, kidneys and endocrine system [1]. The clinical manifestations are very varied among those are: myopathy, exercise intolerance, stroke-like, seizures, ptosis, retinopathy, optic atrophy, blindness, deafness, cardiomyopathy, hepatic and pancreatic diseases, and diabetes. The morphological and biochemical characteristics that are usually associated to mitochondrial myopathy are the presence of ragged-red fibres (RRF) which become evident in muscular biopsies when being tinted with modified trichromic or with the enzymatic reactions of COX, SDH and NADH-TR[2]. Also is necessary to realize studies of breathing and genetic chain study and the presence of lipid accumulations with the tint of oil-red.

Objective: The objective of the work is to describe the discoveries of ultrastructural changes using electron microscopy (ME)

Materials and Methods: In the study were included, patients of the National Institute of Neurology and Neurosurgery of Mexico a total of 342 biopsies were studied with diagnostic of neuromuscular disease. The biopsies with suspicion of Mitochondrial myopathy, including Encephalopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS), Myoclonic epilepsy with ragged red fibres (MERRF), Kearns-Sayre syndrome, progressive external ophthalmoplegia (PEO), Leber's hereditary optic neuropathy (LHON) and others with myopathic manifestations or multisystemic disease not classifiable in described syndrome, were carried out the analysis of electron microscopy. Samples of muscle biopsies were fixed in 0.2% glutaraldehyde and postfixed in 1% OsO₄, dehydrated in acetone and embedded in epon. Ultrathin sections were stained with acetate uranile and citrate of lead. Ultrathin sections were examined with a transmission electron microscope [3].

Results: In 38% (127) of the cases it was found characteristics of mitochondrial myopathy. In the results, mitochondrial accumulation is observed, alterations in its architecture or with the presence of dense inclusions in its interior. Figures (1,2,3).

Conclusions: The variability of the expression of these illnesses hinders its diagnosis, in the event of a clinical suspicion it is necessary to carry out a muscular biopsy, supporting the diagnostic with Electron Microscopy.

References:

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- [2] S. DiMauro. *Biochimica et Biophysica Acta* (2004) 1658, 80.
- [3] V. Dubowitz. *Muscle Biopsy* (1985) 118.

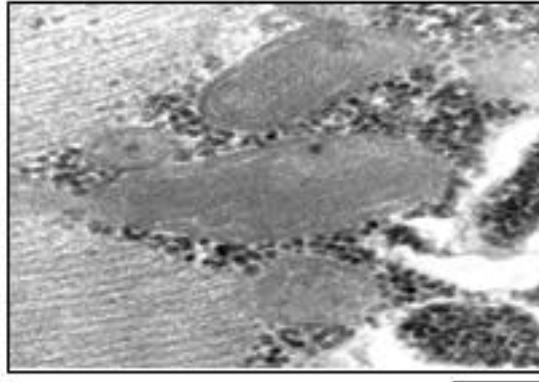


Figure 1. Electron micrography, abnormal mitochondria with paracrystalline inclusion. Bar 250 nm.

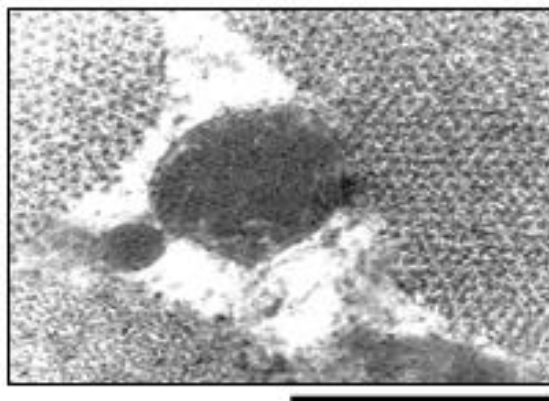


Figure 2. Electron micrography, two mitochondrias containing dense granular inclusion. Bar 250 nm.

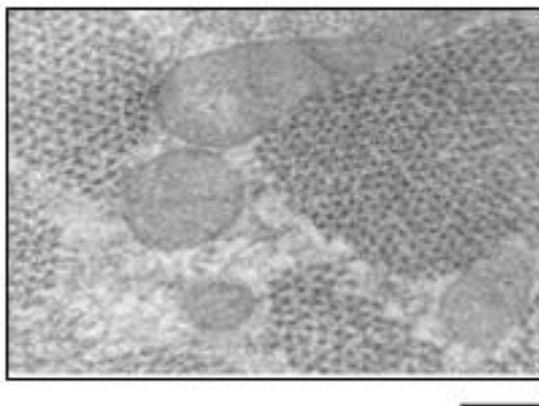


Figure 3. Electron micrography, abnormal mitochondrias with different size and structure. Bar 250 nm.