MITOCHONDRIAL OCULAR MYOPATHY: A CLINICAL, LABORATORY, AND MORPHOLOGIC STUDY (Abstract)*. **Thesis. São Paulo, 1994.**

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We studied 63 patients suffering from mitochondrial ocular myopathy, who were distributed into four groups according to ther clinical manifestations. Patients in Group I presented only bilateral palpebral ptosis; patients in Group II presented palpebral ptosis and bilateral external ophthalmoparesis; patients in Group III had dysphagia and/or dysphonia accompanied by palpebral ptosis and/or bilateral external ophthalmoparesis; patients in Group IV presented a clinical picture of muscular weakness in the upper and lower limbs and face, accompanied by palpebral ptosis and/or bilateral external ophthalmoparesis, with possibility of the occurrence of dysphagia and/or dysphonia.

All cases followed pre-established criteria of inclusion or exclusion with the objective of barring other affections which could present a similar clinical picture. All patients were submitted to several different examinations: clinical with eyegrounds study; routine and specific laboratory tests, such as serum enzyme dosage, T3, T4, TSH, electrocardiography and electromyography. Tests were also done for CSF, thorax and cranium CT and cranium MNR. Lactic acid at rest and after exercise were dosed in 31 cases. All patients underwent muscle biopsy with anatomopathologic study through light microscopy, by means of routine and histochemical techniques and through electronic microscopy.

Two control grousps were assessed. The first one was used for the study of lactic acid, and comprised 20 apparently healthy individuals. The second group comprised 50 patients; of these, 22 had a diagnosis of mitochondrial myopathy without ocular involvement and 28 had a diagnosis of other different myopathies. Diagnosis of these patients were well established by clinical and laboratory tests and by muscle biopsies, which was complemented by electronic microscopy, whose findings were used in the comparative study with our cases.

Results, including statistical analysis, allowed for the following conclusions:

- the clinical and evolutive analysis led us to infer that mitochondrial ocular myopathy has polymorphic clinical features, however of a familial monomorphic clinical stereotypy;
- the predictive analysis of the patients' prognosis is determined by the clinical manifestations; all patients studied presented slow and protruded evolution, thus demonstrating the benign character of mitochondrial ocular myopathy;
- there is no clinical or laboratory correlation with the disease's degree of seriousness;
- increase of lactic acid showed a statiscally significant correlation with the increase of activity in enzymatic reactions for SDH and absence of activity for COX;
- light microscopy is limited and insufficient for diagnostic conclusions; electronic microscopy is fundamental, sensitive and specific for the diagnosis of mitochondrial myopathies;
- through clinical picture, time of evolution, family history and prognosis, we concluded that none of our cases of mitochondrial ocular myopathy developed into the Kearns-Sayre syndrome; therefore the two are distinct affections;
- even in the cases in which clinical involvement is restricted to the ocular extrinsec musculature, there is indication of the anatomopathological study through muscle biopsy of the brachial biceps;
- mitochondrial morphological alterations were similar, in our cases as well as with mitochondriopathies assessed in the control group; thus, these alterations cannot be utilized in the differential diagnosis between the several different forms of mitochondrial diseases.

KEY WORDS: **mitochondrial ocular myopathy, clinics, diagnosis.**

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