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Introduction: MELAS is one of mitochondrial disease characterized by mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes.

Objective: To analyze patients suffering from of MELAS at Clinical Hospital of Federal University of Paraná: clinical, laboratorial, biochemistry and histological findings; mitochondrial DNA (mtDNA) mutations in tRNA^Leu(UUR) gene; and to compare muscle biopsy and molecular analysis of tRNA^Leu(UUR) gene as diagnostic method to MELAS syndrome.

Method: Study of 9 patients with MELAS with correlation between clinical findings, laboratorial data, biochemical, radiological and electrophysiological findings. Muscle biopsies were evaluated mainly by modified Gomori-trichrome (MGT), succinate dehydrogenase (SDH) and cytochrome c oxidase (COX) stains. DNAmt was obtained from muscle biopsy specimen. The tRNA^Leu(UUR) gene was analyzed by PCR/RLFP and direct sequencing.

Results: The onset was before age 15 years in 6 patients. Stroke-like episodes was present in all patients and the others symptoms reported were vomiting, headache, seizures, weakness, dementia, hearing loss, short stature, ocular symptoms, ataxia and facial neuropathy. Blood lactate levels was increased in 8 patients. Brain image study revealed stroke-like pattern in all patients with unilateral lesion in 5 patients and bilateral in 4 patients. Ragged-red fibers (RRF) occurred in MGT (88.8%) and SDH (100%) stains, but the frequency above 2% of RRF was found in 5 patients on MGT stain and in 8 patients on SDH stain. COX stain analysis showed deficient activity one patient. Strongly succinate dehydrogenase-reactive blood vessels (SSV) occurred in 5 patients which frequency ranged from 33.3% to 75% in these cases. The molecular analysis was possible in 6 patients that showed A3243G mutation on mtDNA in 3 patients.

Conclusion: MELAS patients have variations in their clinical manifestation, but the main dysfunctions of MELAS syndrome, as encephalopathy, stroke-like, headache, vomiting and increased lactate levels can be found in all patients. Stroke-like lesions are more common in temporal, occipital and parietal regions. COX deficiency can occur in MELAS patients. RRF presence was increased in SDH than in MGT stain. Absent SSV in muscle biopsy specimens should not be used as exclusion criteria for MELAS. A3243G point mutation is the most related with the MELAS phenotypic in tRNA^Leu(UUR) gene. Muscle biopsy as diagnostic method is better than molecular analysis of tRNA^Leu(UUR) gene in MELAS syndrome.

Key words: human T-lymphotropic virus 1, major depression, asymptomatic and HAM/TSP HTLV-1.


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