

CLINICAL PRESENTATION AND PREDICTORS OF OUTCOME IN ADULT PATIENTS WITH MITOCHONDRIAL DISEASES

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Introduction: Mitochondrial diseases have diverse clinical, biochemical and genetic presentations. Patients can have a “classic” mitochondrial syndrome or a more heterogeneous phenotype, leading to a challenging prediction of the clinical course and long-term outcome.

Aim: To describe the clinical presentations in a cohort of adults with mitochondrial disease and determine predictors of their outcome.

Methods: Demographics and clinical data were collected for all patients with a probable or definite diagnosis of mitochondrial disease according to the Bernier criteria. Outcomes were defined as death related to complications of the disease and degree of dependency, evaluated with the Katz Index of Independence in Activities of Daily Living.

Results: 60 patients were included, 57% had an adult-onset form and the majority were female (62%). The most common phenotype was chronic progressive external ophthalmoplegia (46%). 6 patients died within follow-up (10%). Death was significantly associated with cardiac involvement ($p=0.03$). The mean Katz score was 5. Patients with a younger age at onset ($p<0.05$), presence of generalized myopathy ($p=0.002$), developmental delay ($p<0.005$) or cardiac involvement ($p<0.05$) as well as a history of encephalopathy ($p<0.01$), epilepsy ($p<0.01$) or stroke-like episodes ($p=0.03$) had a higher degree of dependency (defined as Katz score ≤ 2).

Conclusions: Our patients had mainly mild presentations with ocular involvement. The presence of other clinical findings is important for the degree of dependency and outcome. Cardiac involvement seems to be the main clinical factor related to death in mitochondrial disease, highlighting the importance of systematic cardiac study in these patients.