

## Publication

### Are patients affected by mitochondrial disorders at nutritional risk?

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Patients with mitochondrial disorders (MD) frequently present with gastrointestinal complaints, mainly gastrointestinal dysmotility, that interfere with their food intake. A deterioration of their nutritional state may worsen the course of the disease. Our study aimed to evaluate a simple screening tool to identify nutritional risk and perform an extended nutritional assessment to explore the potential presence of deficiencies in this population compared with controls.; A prospective cohort study was conducted to compare outpatients with MD to matched healthy controls. Nutritional screening and full nutritional assessments were performed, including quantitative and qualitative dietary habits (7-d food log), body function and composition, and resting energy expenditure and quality of life (QoL) measurements. Blood and 24-h urine sample analyses were performed in the patient group.; Twenty-six subjects were included in the study, with 11 in the patient group and 15 in the control group. No patient was deemed malnourished according to the nutritional risk score NRS-2002. When compared with the controls, however, the patients with MD had significantly lower muscle mass ( $P=0.04$ ), reduced handgrip strength ( $P=0.07$ ), and significant changes in QoL and pathologic creatinine height index, which indicate malnutrition. The patients with MD also had a significantly lower protein intake ( $P=0.01$ ).; According to the current definition by the European Society of Clinical Nutrition and Metabolism (ESPEN) and the American Society of Parenteral and Enteral Nutrition (ASPEN), all patients fulfilled the criteria for malnutrition. Thus, the usual nutritional screening tool is less sensitive for chronically ill outpatients. These results provide a rationale to increase protein intake and adapt patients' energy stores to improve symptoms and QoL.

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