RIBOFLAVIN IS INEFFECTIVE FOR MATERNALLY INHERITED MITOCHONDRIAL DISEASE

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Introduction

With interest we read the article by Darin et al. about 13 members of a family carrying the m.3250T>C mutation in the tRNA(Leu) gene on the mitochondrial DNA. Two of these patients were systematically treated with riboflavin but without a beneficial effect. We have the following comments and concerns.

The rationale for treating two patients (II/2, II/5) with a mitochondrial disorder (MID) with riboflavin is unclear. From vitamins, co-factors and antioxidants it is well-known that they are usually ineffective in MIDs. This is particularly the case for riboflavin. There are no systematic and controlled investigations available testing the effect of riboflavin in patients with MIDs. Only in some cases was a questionable beneficial effect attributed to riboflavin.

Coenzyme-Q or its derivatives have been shown beneficial in single cases particularly in those with primary coenzyme-Q deficiency. Why did only two patients receive coenzyme-Q?

All 13 patients had tachycardia or tachypnoea on exertion (table 1). However, in 8 patients the routine ECG was normal at least at two recordings. How do the authors explain this discrepancy? Did these patients receive anti-arrhythmic medication for tachycardia? Was it sinustachycardia, atrial tachycardia, or ventricular tachycardia?

If a patient can walk for 20km (patient III/7) why does he need a walker? Did those who were able to walk for >3km with a walker need the device because of ataxia, vertigo, or sensory disturbance? Seven patients were able to run 200 to 1000m. Why do patients who are able to run need a walking aid?

Patients with exertional tachycardia or tachypnoea not only require clinical cardiologic exam, blood pressure measurement, and ECG but also transthoracic echocardiography and thoracic ultrasound. Did the 13 patients undergo these examinations and which were the results? In how many was exertional dyspnoe attributable to heart failure or systolic dysfunction, and in how many to pulmonary disease?

All 13 patients had muscle symptoms upon exercise or even at rest? How do the authors explain that despite the presence of myopathy creatine-kinase (CK) was elevated in only 5 patients? Was CK measured at rest or after exercise?

Overall, this interesting case series requires more extensive clinical and instrumental examinations. Riboflavin should not be given to patients with a MID.

References

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