Human Coenzyme Q_{10} Deficiency

Catarina M. Quinzii · Salvatore DiMauro · Michio Hirano

Accepted: 3 October 2006 / Published online: 10 November 2006

Abstract Ubiquinone (coenzyme Q_{10} or CoQ_{10}) is a lipid-soluble component of virtually all cell membranes and has multiple metabolic functions. Deficiency of CoQ_{10} (MIM 607426) has been associated with five different clinical presentations that suggest genetic heterogeneity, which may be related to the multiple steps in CoQ_{10} biosynthesis. Patients with all forms of CoQ_{10} deficiency have shown clinical improvements after initiating oral CoQ_{10} supplementation. Thus, early diagnosis is of critical importance in the management of these patients. This year, the first molecular defect causing the infantile form of primary human CoQ_{10} deficiency has been reported. The availability of genetic testing will allow for a better understanding of the pathogenesis of this disease and early initiation of therapy (even presymptomatically in siblings of patients) in this otherwise life-threatening infantile encephalomyopathy.

Keywords CoQ10 · Mitochondria · Encephalopathy · Myopathy · Cerebellar ataxia