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Titel des Beitrags:
Congenital cataract, muscular hypotonia, developmental delay and sensorineural hearing loss associated with a defect in copper metabolism.

Abstract:
Deficiencies of different proteins involved in copper metabolism have been reported to cause human diseases. Well-known syndromes, for example, are Menkes and Wilson diseases. Here we report a patient presenting with congenital cataract, severe muscular hypotonia, developmental delay, sensorineural hearing loss and cytochrome-c oxidase deficiency with repeatedly low copper and ceruloplasmin levels. These findings were suggestive of a copper metabolism disorder. In support of this, the patient's fibroblasts showed an increased copper uptake with normal retention. Detailed follow-up examinations were performed. Immunoblotting for several proteins including ATP7A (MNK or Menkes protein), ATP7B (Wilson protein) and SOD1 showed normal results, implying a copper metabolism defect other than Wilson or Menkes disease. Sequence analysis of ATOX1 and genes coding for proteins that are known to play a role in the mitochondrial copper metabolism (COI-III, SCO1, SCO2, COX11, COX17, COX19) revealed no mutations. Additional disease genes that have been associated with cytochrome-c oxidase deficiency were negative for mutations as well. As beneficial effects of copper histidinate supplementation have been reported in selected disorders of copper metabolism presenting with low serum
copper and ceruloplasmin levels, we initiated a copper histidinate supplementation. Remarkable improvement of clinical symptoms was observed, with complete restoration of cytochrome-c oxidase activity in skeletal muscle.