term antibacterial therapy, careful follow-up and monitoring with spinal x-ray and neurologic evaluation, as indicated in the Great Ormond Street experience of spinal TB.

METABOLIC DISORDERS

CLINICAL CHARACTERISTICS OF 5 PHENOTYPES OF COENZYME Q10 DEFICIENCY

Researchers at Columbia University Medical Center, New York; University of Genoa, Italy; and University of Granada, Spain reviewed 149 cases of coenzyme Q10 (ubiquinone) deficiency, including their own cohort of 76 patients diagnosed from 1997-2010. Cerebellar ataxia was the principal phenotype and the presenting symptom in 94 children (63%). Less frequent phenotypes included encephalomyopathy in 4 patients, isolated myopathy in 14, infantile-onset multisystemic disease in 17, nephropathy (with or without sensorineural hearing loss) in 11, and atypical presentations in 9. Other manifestations include neuropathy, seizures, congenital hypotonia, dystonia, ophthalmoplegia, retinitis pigmentosa, optic atrophy, agenesis of corpus callosum, and hypogonadism. Onset was primarily in childhood; 82% were aged < 13 years including 23% in infancy (<12 months). Mortality rate was 8%.

Direct measurement of CoQ10 in skeletal muscle by liquid chromatography is the most reliable test for diagnosis of CoQ10 deficiency. Morphological and biochemical findings differ in the various clinical forms. Family history suggests autosomal recessive inheritance. Pathogenic mutations are described in patients with the infantile multisystemic syndrome and some juvenile-onset cerebellar ataxia cases. Response to oral supplementation with CoQ10 is frequent but variable; one patient with infantile spasms failed to respond. (Emmanuele V, Lopez LC, Berardo A, et al. Heterogeneity of coenzyme Q10 deficiency. Patient study and literature review. **Arch Neurol** 2012 Aug;69(8):978-83). (Respond: Michio Hirano MD, H Houston Merritt Clinical Research Center, Department of Neurology, Columbia University Medical Center, 630 W 168th St, P&S 4-423, New York, NY 10032. E-mail: mh29@columbia.edu).

COMMENT. The occurrence of primary and secondary CoQ10 deficiencies adds to the difficulty in study of the molecular classification of this heterogeneous disorder. (Quinzii CM, Hirano M. **Biofactors** 2011 Sep;37(5):361-5). Pathogenic mutations are identified in genes involved in the biosynthesis of CoQ10 (primary CoQ10 deficiencies) or in genes not directly related to CoQ10 biosynthesis (secondary CoQ10 deficiencies). Respiratory chain defects may contribute to the pathogenesis of primary CoQ10 deficiencies.

HEADACHE DISORDERS

MANIFESTATIONS OF FAMILIAL HEMIPLEGIC MIGRAINE

Researchers at University of Arkansas, Little Rock, AR report 3 cases of familial hemiplegic migraine complicated by reversible cerebral edema and followed by