METABOLIC AND DEGENERATIVE DISORDERS

ALTERNATING HEMIPLEGIA: MITOCHONDRIAL DYSFUNCTION

Mitochondrial function in 4 patients with alternating hemiplegia (AH) was evaluated by magnetic resonance spectroscopy of resting muscle at the Montreal Neurological Institute, McGill University, Montreal, Canada. All patients had abnormally high resonance intensities from inorganic phosphate and an abnormally low calculated cytosolic phosphorylation potential. These changes were consistent with mitochondrial dysfunction. (Arnold DL et al. Evidence for mitochondrial dysfunction in patients with alternating hemiplegia of childhood. Ann Neurol June 1993; 33: 604-607). (Respond: Dr Arnold, Montreal Neurological Institute, 3801 University Street, Montreal, Quebec, H3A 2B4 Canada).

> COMMENT. Alternating hemiplegia of childhood may represent a phenotype of mitochondrial disease. Mitochondrial diseases share with AH the occurrence of alternating hemiplegia, seizures, and neurodevelopmental deterioration. The clinical characteristics and differential diagnosis of AH are reviewed in a report of 22 cases from the Hopital des Enfants Malades, Paris, France. (Bourgeois M et al. J Pediatr May 1993; 122: 673-9). SPECT images of two patients showed focal areas of decreased uptake of the radiotracer, representing impaired regional blood flow during and between hemiplegic episodes and a possible mechanism of the neurologic deficits.(Siemes H, Cordes M. Dev Med & Child Neur April 1993; 35: 346).

RETT SYNDROME: A MITOCHONDRIAL DYSFUNCTION?

Ultrastructural and biochemical alterations of muscle mitochondria are reported in two girls, aged 4 years, with Rett syndrome examined at the Institute of Neurological Sciences, University of Siena, Italy. Mitochondria were abnormally swollen and dumb-bell-shaped, and cytochrome c oxidase and NADH cytochrome c reductase were decreased in activity. The primary or secondary role of this mitochondrial pathology in the pathogenesis of the syndrome is undetermined. (Dotti MT et al. Mitochondrial dysfunction in Rett syndrome. Brain & Development March/April 1993; 15: 103-106). (Respond: Prof A Federico, Instituto di Scienze Neurologiche, Policlinico Le Scotte, 53100 Siena, Italy).

> COMMENT. In contrast to the above, phosphorus magnetic resonance spectroscopy, proton MRS, muscle biopsies, and determination of pyruvate and lactate in plasma of 5 girls with Rett syndrome, examined at the John F Kennedy Institute, Glostrup, Denmark, failed to detect evidence for a mitochondrial disorder. (Nielsen JB, Lou HC et al. Brain & Development March/April 1993; 15: 107-112).