

for RS.

Four cases of RS with a single family tree and prevalence rate of 2.1 per 10,000 are reported from Northern Tuscany, Italy. (Pini G, Milan M, Zappella M. Clin Genet Dec 1996;50:486-490). This study supports a genetic basis for RS.

METABOLIC DISORDERS

SUBEPENDYMAL CYSTS AND BIOTIN DEFICIENCY

Subependymal cysts were identified by cranial ultrasound and confirmed by MRI in an infant with holocarboxylase synthetase (HCS) deficiency, presenting with lactic acidosis, shock, and hypertonia, and responsive to biotin therapy (10mg daily), in a report from Devos Children's Hospital, Grand Rapids, MI. At delivery the infant had apnea and bradycardia, and Apgars were 7 and 8. Within hours she developed tachypnea, fever, and shock. Laboratory studies showed metabolic acidosis, hypoglycemia, lactic acidosis, and elevated pyruvate and ammonia. Urinary organic acid assays revealed increased 3-hydroxyisovaleric acid, 3-methyl-crotonoylglycine, and methylcitric acid, diagnostic of multiple carboxylase deficiency. Treatment with bicarbonate and biotin was followed by remission of symptoms and a normal development at 14 month follow-up. MRI at age 6 months showed resolution of the bilateral subependymal cysts and normal myelination. (Squires L, Betz B, Umfleet J, Kelley R. Resolution of subependymal cysts in neonatal holocarboxylase synthetase deficiency. Dev Med Child Neurol April 1997;39:267-269). (Respond: Liza Squires MD, Pediatric Neurology, Devos Children's Hospital, 330 Barclay NE, Grand Rapids, MI).

COMMENT. Subependymal cysts uncovered by cranial ultrasound in a sick neonate require investigation of possible metabolic disorders. Prompt diagnosis and specific therapy can prevent fatalities and permit normal development.

Glutaric aciduria type 1 is cited in association with cerebral arachnoid cysts (Millichap JG. Neurology 1997;48:1435); and L-2-hydroxyglutaric aciduria is reported in 6 Portuguese children presenting with mental deficiency, cerebellar ataxia, progressive macrocephaly and seizures. (Barbot C, Fineza I, Diogo L et al. L-2-hydroxyglutaric aciduria: clinical, biochemical and magnetic resonance imaging in six Portuguese pediatric patients. Brain Dev June 1997;19:268-273). A thalamic tumor, a diffuse fibrillary astrocytoma, was found in one of these cases, the second in the literature described with hydroxyglutaric aciduria.

MITOCHONDRIAL ENCEPHALOPATHY AND CYTOCHROME C

Benefits from treatment of mitochondrial encephalomyopathy (MEM) with cytochrome C (6.25 mg) and vitamins B1 (25 mg) and B2 (12.5 mg), in daily injections, are reported from Osaka University Medical School and other centers in Japan. Symptomatic improvements in 8 of 9 patients included decrease in muscle fatigability, and lessening of motor disability and severity of stroke-like episodes. Intermittent courses of injections were needed to maintain clinical improvement. (Tanaka J, Nagai T, Arai H et al. Treatment of mitochondrial encephalomyopathy with a combination of cytochrome C and vitamins B1 and B2. Brain Dev June 1997;19:262-267). (Respond: Dr Junko Tanaka, Sakai Municipal Hospital, Minamiasui-cho 1-1-1, Sakai, Osaka 590, Japan).

COMMENT. The authors advocate a trial of cytochrome C/vitamin B treatment in children with MEM; disease progression is not halted but symptoms may be ameliorated.

BONE MARROW TRANSPLANT IN ADRENOLEUKODYSTROPHY

Three children with adrenoleukodystrophy (ALD) were treated with allogeneic bone marrow transplantation (BMT) at Huddinge University Hospital, Sweden. Patient 1, a boy aged 9, presented with attention deficit disorder and learning disability; MRI showed demyelination in the frontal lobes which progressed over a 4 year follow-up, in association with mental deterioration. The 2nd and 3rd patients had no symptoms but white matter lesions were present on MRI at diagnosis. One had Addison's disease at 6 years of age, and ALD was confirmed biochemically and by MRI six months later. He showed worsening of attention deficits before, and he died 18 months after BMT. Patient 3 was the younger brother of patient 2, and ALD and Addison's disease were diagnosed at 4 years. This boy is healthy 3.5 years after BMT. (Malm G, Ringden O, Anvret M et al. Treatment of adrenoleukodystrophy with bone marrow transplantation. Acta Paediatr May 1997;86:484-492). (Respond: Dr G Malm, Department of Paediatrics, Huddinge University Hospital, 14186 Huddinge, Sweden).

COMMENT. The authors recommend bone marrow transplantation implemented early in the treatment of ALD, even without symptoms, provided that the MRI shows signs of demyelination and a suitable donor is available.

Attention deficit disorder was the presenting clinical manifestation of ALD in one of these patients, emphasizing the neurologist's role in the management of children with ADHD.

HYPOGLYCEMIA AND COGNITIVE DEFICITS

Effects of hypoglycemia on cognition were studied using event-related brain-potential (ERP) measures and reaction times at the Otto-von-Guericke University, Magdeburg, Germany. ERP measures of selective attention, response choice and reaction time were delayed during hypoglycemia compared to baseline performance. After restoration of euglycemia, total-error frequencies were still higher than at baseline and recovery of the quality of task performance was delayed. Hypoglycemia impairs both the stimulus and the motor-response selection. Large negative shifts in cortical potentials were distributed over frontal areas during hypoglycemia, suggesting that the frontal cortex is involved in the control of attention and stimulus selection. (Smid HGOM, Trumper BG, Pottag G et al. Differentiation of hypoglycaemia induced cognitive impairments. An electrophysiological approach. Brain June 1997;120:1041-1056). (Respond: Dr Smid, Neurophysiology Clinic, Otto-von-Guericke University, Leipzigerstrasse 44, 39120, Magdeburg, Germany).

COMMENT. Hypoglycemia has an adverse effect on attentional behavior by increasing reaction times and error frequencies. After restoration of normal blood sugar levels there is a delay in recovery of quality of task performance. The frontal cortex is more highly activated during acute hypoglycemia as a compensatory mechanism for the reduced energy supply to the brain. Frontal location brain mechanisms, important in control of attention, are particularly affected by the impaired energy supply associated with hypoglycemia.