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, Minamiyasui-cho 1-1-1, Sakai, Osaka 590, Japan).