

Aug;49(2):130-133.e1). (Response: Dr Bain, UMDNJ – New Jersey Medical School; Pediatrics: 185 S Orange Ave, Newark, NJ 07101. E-mail: jenniferbain08@gmail.com).

COMMENT. Reversible cerebral vasoconstriction syndrome, sometimes called Call-Fleming syndrome (Call GK, et al. **Stroke** 1988 Sep;19(9):1159-70) is rare in children but should be considered in the differential diagnosis of a child with a history of migraine and presentation with thunderclap headache. Many risk factors in addition to migraine are listed, including vasoactive drugs (triptans, amphetamines, ginseng, nicotine patch, oral contraceptives, diet pills, selective serotonin reuptake inhibitors), blood products, and trauma. The disorder is self-limiting, the headache resolving in 3 weeks and vasoconstriction resolving by 12 weeks. The majority has no residual symptoms and no recurrence of headaches, although stroke is a potential complication. Children treated with methylphenidate or amphetamines for ADHD and having a history of headache should be followed closely and the stimulant discontinued if headaches are persistent.

DEVELOPMENTAL DISORDERS

CORPUS CALLOSUM IN SMITH-LEMLI-OPITZ SYNDROME

Investigators at the Kennedy Krieger Institute and Johns Hopkins University, Baltimore; and the National Institutes of Health, Bethesda, MD, studied the relation between the size of the corpus callosum (CC), level of serum 7-dehydrocholesterol (7DHC), and severity of developmental delay in 36 subjects with Smith-Lemli-Opitz syndrome (SLOS), compared to 36 normal controls. The mean age was 3.9 yrs (range 0.2 – 12.5 yrs); 18 boys and 18 girls. Shorter CC length measured on one midsagittal image and smaller CC area correlated with a lower developmental quotient in gross motor and language domains, ranging from mild to severe, but not with fine motor or adaptive skills. Also, CC length and area negatively correlated with the 7DHC and 8DHC levels, and positively correlated with serum total cholesterol level. (Lee RW, Yoshida S, Jung ES, Mori S, Baker EH, Porter FD. Corpus callosum measurements correlate with developmental delay in Smith-Lemli-Opitz syndrome. **Pediatr Neurol** 2013 Aug;49(2):107-12). (Response: Dr Lee, Kennedy Krieger Institute, Baltimore, MD 21205. E-mail: leer@kennedykrieger.org).

COMMENT. SLOS (or 7-dehydrocholesterol reductase deficiency) is an autosomal recessive metabolic and developmental congenital disorder, first described in 1964 (Smith DW, Lemli L, Opitz JM. A newly recognized syndrome of multiple congenital anomalies. **J Pediatr** 1964 Feb;64:210-7). The most commonly observed features include dysmorphic faces, microcephaly, 2-3 syndactyly of toes, polydactyly, growth retardation, intellectual disability, cleft palate, and hypospadias. Hypoplasia/agenesis of the corpus callosum is reported less frequently, as well as holoprosencephaly. Mutations in the DHCR7 gene are the cause, the enzyme responsible for production of cholesterol, an essential nutrient for embryonic development. Sequencing analysis of DHCR7 detects ~96% of known mutations. (Irons M. Smith-Lemli-Opitz Syndrome. In: Pagon RA, Adam MP, Bird TD, et al., editors. **GeneReviews** [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013).

MRI and 1H-MRS may prove effective in assessment of effects of cholesterol replacement therapy in patients with SLOS (Caruso PA et al. **Neuroradiology** 2004 Jan;46(1):3-14). Of 18 patients with SLOS, abnormal CNS findings were noted in 5 patients, including callosal abnormalities in 4 (22%), Dandy-Walker variant in 1, arachnoid cyst in 1, and holoprosencephaly in 1 (6%). Clinical degree of disease severity was correlated with lipid:choline ratios.

DEVELOPMENTAL OUTCOME FOLLOWING HEMISPHERECTOMY FOR HEMIMEGALENCEPHALY

Investigators at the National Center of Neurology and Psychiatry, Tokyo, studied the effect of early hemispherectomy on development in a consecutive series of 12 infants with hemimegalencephaly (HME) and epileptic encephalopathy. Mean age at onset was 20.4 days, mean age at surgery was 4.3 months (range 2-9), and mean follow-up time was 78.8 months (36-121). Eleven patients had a history of early infantile epileptic encephalopathy. Following vertical parasagittal hemispherectomy, 8 (66.7%) were seizure-free and showed significantly higher postoperative developmental quotient (DQ) than those with seizures (mean 31.3 vs 5.5; $p=0.02$). In the seizure-free group, postoperative DQ correlated with postoperative seizure duration ($p=0.01$). Shorter seizure duration during early infancy provides better postoperative DQ in infants with HME and epileptic encephalopathy. (Honda R, Kaido T, Sugai K, et al. Long-term developmental outcome after early hemispherectomy for hemimegalencephaly in infants with epileptic encephalopathy. **Epilepsy Behav** 2013 Aug 6;29(1):30-35). (Response: Dr Kaido. E-mail: kaido@ncnp.go.jp).

COMMENT. Despite the risks, the authors conclude that early surgical intervention for hemimegalencephaly is preferable to brain damage from repeated seizures and encephalopathy and long-term AED use. A similar conclusion was reached by investigators at the Sorbonne, Paris, France, who reviewed the literature and reports of ~600 cases in the last 30 years (Bulteau C et al. Epilepsy surgery for hemispheric syndromes in infants: hemimegalencephaly and hemispheric cortical dysplasia. **Brain Dev** 2013 Sep;35(8):742-7). Hemispheric surgical procedures are considered safe and can be performed from the first month of life. Residual insular cortex and contralateral MRI abnormalities are associated with persistent postoperative seizures and lack of cognitive improvement.

NEURO CUTANEOUS DISORDERS

mTOR INHIBITION AND TUBEROUS SCLEROSIS PREVENTION

Investigators at Children's Memorial Health Institute, Warsaw, Poland, report monozygotic twin sisters with tuberous sclerosis complex (TSC), one treated with the mTOR inhibitor everolimus since age 4 years. After 24-month follow-up, everolimus treatment was associated with a significant decrease in brain tumor volume, and the treated twin presents no facial angiofibroma, and no renal angiomyolipomas (AMLs).