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COMMENT. Posterior reversible encephalopathy syndrome (PRES), first described in 1996 (Hinchey J, et al. **N Engl J Med** 1996 Feb 22;334(8):494-500), is characterized clinically by headache, altered awareness, visual disturbance, and seizures, and radiologically by transient posterior lesions in subcortical white matter. PRES is associated with a rapid rise in blood pressure that may underlie the encephalopathy. The pathophysiology of PRES is not completely understood, but predisposing conditions include renal and hemato-oncologic diseases and use of chemotherapeutic immunosuppressive drugs. (Siebert E, et al. **Eur J Paediatr Neurol** 2013 Mar;17(2):169-75 [Cited by Mameli]). Other conditions reported in association with PRES are organ and bone marrow transplantation, autoimmune disease, Guillain-Barre syndrome, sickle cell anemia, hemolytic-uremic syndrome, and iv immunoglobulin administration.

***SLC19A3* EARLY-INFANTILE, LETHAL ENCEPHALOPATHY**

Investigators from VU Medical Centre, Amsterdam, The Netherlands, identified seven patients with severe encephalopathy who shared a previously undescribed MRI pattern with cystic degeneration of the white matter and progressive cerebral, cerebellar and brainstem atrophy. All patients showed rapid deterioration of brain function soon after birth, followed by respiratory failure and death. Whole-exome sequencing revealed pathogenic, heterozygous missense mutations in the *SLC19A3* gene, encoding the second thiamine transporter. Pathology of brain tissue demonstrates cerebral atrophy and lesions similar to Leigh's syndrome. This new, severe, lethal phenotype broadens the phenotypic spectrum of *SLC19A3* mutations and is recognized by the associated MRI pattern of brain degeneration. (Kevelam SH, Bugiani M, Salomons GS, et al. Exome sequencing reveals mutated *SLC19A3* in patients with an early-infantile, lethal encephalopathy. **Brain** 2013 May;136(Pt 5):1534-43). (Response: Marjo S van der Knaap, Department of Child Neurology, VU Medical Centre, de Boelelaan 1117, 1081 HV Amsterdam, The Netherlands. E-mail: ms.vanderknaap@vumc.nl).

COMMENT. MRI pattern of initial swelling with T-hyperintensities followed by rapid degeneration and brain atrophy allows early diagnosis of a rapidly progressive infantile encephalopathy caused by *SLC19A3* mutations.

MIGRATING PARTIAL SEIZURES OF INFANCY

A national surveillance study in conjunction with the British Paediatric Neurology Unit was undertaken to further define the clinical, pathological and molecular genetic features of migrating partial seizures of infancy (MPSI), a rare early infantile epileptic encephalopathy with poor prognosis. In 14 patients reported during the 2 year study period, MPSI was associated with an expanded spectrum of clinical features including gut dysmotility and movement disorder, EEG features including hypersarrhythmia with infantile spasms and burst suppression, and novel brain imaging including delayed myelination, white matter hyperintensity and in one patient at autopsy, putaminal

atrophy. Two further autopsied cases showed hippocampal gliosis and neuronal loss. Two patients had mutations in the KCNT1 gene, while genetic testing for other known early infantile epileptic encephalopathy genes (including PLCB1 and SLC25A22) was negative. (McTague A, Appleton R, Avula S, et al. Migrating partial seizures of infancy: expansion of the electroclinical, radiological and pathological disease spectrum. **Brain** 2013 May;136(Pt 5):1578-91). (Response: Dr R Kneen, University of Liverpool. E-mail: rachel.kneen@liverpool.ac.uk).

COMMENT. Investigators at the Children's Hospital Boston found that loss of PLCB1 function is one cause of malignant migrating partial seizures in infancy (MMPEI), but screening of further cases for PLCB1 deletions or mutations was negative. (Poduri SA, et al. **Epilepsia** 2012 Aug;53(8):e146-50).

Investigators at University of Melbourne, Australia, screened 15 unrelated children with migrating partial seizures of infancy (MPSI) for mutations in several genes associated with infantile epileptic encephalopathies. One patient had a de novo SCN1A missense mutation, and MPSI is the most severe SCN1A phenotype to-date. Epilepsies associated with SCN1A mutations range in severity from febrile seizures to severe epileptic encephalopathies including Dravet syndrome and severe infantile multifocal epilepsy. (Carranza RD, et al. **Neurology** 2011 Jul 26;77(4):380-3).

Nordli DR at Lurie Children's Hospital of Chicago, in discussing epileptic encephalopathies in infants and children, notes that similar gene mutations have been found in several different epilepsy syndromes, and accurate classification of these severe epilepsies is important as the first step toward improved treatment and outcome. (Nordli DR Jr. **J Clin Neurophysiol** 2012 Oct;29(5):420-4).

A complex V *ATP5A1* defect causes fatal neonatal mitochondrial encephalopathy in two siblings reported from Radboud University Medical Centre, Nijmegen Centre for Mitochondrial Disorders, The Netherlands. (Jonckheere AI, Renkema GH, Bras M, et al. **Brain** 2013 May;136(Pt 5):1544-54). Exome sequencing revealed a heterozygous mutation in the *ATP5A1* gene.

***ADORA2A* polymorphism predisposes children to encephalopathy** with febrile status epilepticus in a study of 85 patients with acute encephalopathy. AA diplotype of *ADORA2A* is associated with a higher risk of developing seizures and excitotoxic brain damage. (Shinohara M, Saitoh M, Nishizawa D, et al. **Neurology** 2013 Apr 23;80(17):1571-1576).

INFECTIOUS DISORDERS

ACUTE CEREBELLAR ATAXIA AND LYME DISEASE

Child neurologists at Baskent University Faculty of Medicine, Turkey, report the case of a 5-year-old girl from the Mediterranean region of Anatolia with a 4-day history of progressive ataxia. History of fever, rash or tick bite was absent. Neurologic examination revealed cerebellar signs without signs of meningitis or cranial nerve involvement. CT and MRI were normal and CSF showed a mild pleocytosis and normal protein and glucose. Serological evaluation for *Borrelia burgdorferi* was positive and IV