Department of Pediatrics, L Sacco Hospital, University of Milan, 20157 Milano, Italy. E-mail: mameli.chiara@hsacco.it).

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 hypsarrhythmia with infantile spasms and burst suppression, and novel brain imaging including delayed myelination, white matter hyperintensity and in one patient at autopsy, putaminal