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

cefotaxime was begun. Serum markers for other infectious diseases sometimes complicated by cerebellar ataxia were negative; these included herpes simplex, cytomegalovirus, varicella zoster, mumps, rubella, rubeola, Epstein-Barr virus, and mycoplasma. At discharge on day 28, the neurologic exam was normal, and serum for B burgdorferi IgM and IgG antibodies was positive. (Erol I, Saygi S, Alehan F. Acute cerebellar ataxia in a pediatric case of Lyme disease and a review. **Pediatr Neurol** 2013 May;48(5):407-10). (Resp: Dr Erol, Adana, Turkey. E-mail: ilknur_erol@yahoo.com).

COMMENT. Neuroborreliosis presents with both central and peripheral nervous system manifestations, including aseptic meningitis, meningoencephalitis, Bell's palsy, radiculoneuritis, and myelitis. Four previously published reports of cerebellar ataxia with Lyme disease are reviewed.

MRI AS ADJUNCT IN DIAGNOSIS OF MENINGITIS

Investigators from the Children's Hospital of Pittsburgh, PA, reviewed the literature on the role of MRI as an adjunct for diagnosing meningitis. Of 7 relevant articles, two were reviews and an opinion of usefulness of the MRI was based on 5 articles. Specificity of MRI (i.e. negative imaging findings in those who did not have meningitis) was high and ranged from 93% to 100%. Sensitivity of the MRI was more variable (9%, 85%, 95% and 100%); sensitivity may be higher for bacterial and fungal meningitis than for viral meningitis, but it may depend on the degree of inflammatory response and may vary with etiology. The MRI sequences may vary in yield, the contrast-enhanced FLAIR being most useful in a number of studies. Most of the studies included children, but the majority involved adults.

Based on the studies reviewed, MRI is not recommended to rule out meningitis due to its poor sensitivity; it may be more useful for bacterial compared to viral meningitis, but sensitivity varies depending on MRI technique used and data are limited. MRI is more specific but cannot be recommended to rule in meningitis because data are limited for children and none for infants. (Upadhyayula S. Is there a role for MRI as an adjunct for diagnosing bacterial meningitis? **Arch Dis Child** 2013 May;98(5):388-90). (Response: Dr Shankar Upadhyayula, Infectious Diseases, Children's Hospital of Pittsburgh, PA 15206. E-mail: Shankar.upadhyayula@chp.edu).

COMMENT. If further studies provide a more definitive role, MRI could be of diagnostic value in neonates and small children with traumatic lumbar punctures, to avoid unnecessary long-term antibiotics and extended hospital stay.

NEUROCUTANEOUS DISORDERS

GM1 GANGLIOSIDOSIS TYPE 1 AND MONGOLIAN SPOTS

Investigators in Sao Paulo, Brazil, report a female infant born at term to healthy consanguineous parents who was examined at 9 months for delayed development. She showed hepatosplenomegaly, and widespread Mongolian spots extending over the back