

posterior white matter hyperintensities and mild cerebellar vermis atrophy, and the muscle biopsy showed vacuolization of the sarcotubular system. EEG was normal in 2 patients and showed epileptiform discharges in 2, generalized in one and bioccipital in another. The defective gene was localized to a new disease locus on chromosome 16q21-q23. Epilepsy presented at age 9-12 months, and ataxia was noted when the children started to walk at 2-3 years. All 4 had psychomotor delay and learning disabilities. Deep tendon reflexes were diminished, plantar responses were equivocal, speech was dysarthric, and the eye exam showed nystagmus. (Gribaa M, Salih M, Anheim M et al. A new form of childhood onset, autosomal recessive spinocerebellar ataxia and epilepsy is localized at 16q21-q23. **Brain** July 2007;130:1921-1928). (Respond: Dr M Koenig, Institut de Genetique et de Biologie Molculaire et Cellulaire, 1 rue Laurent Fries BP10142, 67404 Illkirch cedex, France).

COMMENT. The differential diagnosis of recessive spinocerebellar ataxia with progressive myoclonus epilepsy and/or generalized tonic-clonic seizures includes Unverricht-Lundborg disease, Lafora disease, neuronal ceroid lipofuscinoses, sialidoses, the sensory ataxia, neuropathy, dysarthria and ophthalmoparesis (SANDO) syndrome, and myoclonic epilepsy with ragged red fibers (MERRF) syndrome. Age at onset, absence of myoclonus and dementia, and EEG and MRI findings allowed exclusion of these disorders and the definition of a new epilepsy/ataxia syndrome localized to the 16q21-q23 locus.

DIAGNOSTIC INACCURACIES IN CHILDREN WITH “FIRST SEIZURE”

A prospective cohort study of 127 children aged 1 month through 17 years seen in the First Seizure clinic at the Alberta Children's Hospital between Jan 1, 2004 and August 30, 2005 determined the range of diagnoses and the prevalence of previous unrecognized seizures. The diagnosis was epileptic in 94 (74%), nonepileptic in 31 (24%) and unclassified in two (2%). Pediatricians referred true epileptic events in 92% cases, ED physicians 76%, and family physicians 65%. Mean age at presentation was 8 years. Development was delayed in 15%; the neurologic examination was abnormal in 11%. True epileptic seizures were generalized in 32 (34%) and partial in 62 (66%). Fifteen (16%) had an epilepsy syndrome. Over a 1-year follow-up period, 42 (45%) children presenting with an epileptic seizure were diagnosed with epilepsy (recurrent seizures). A prior probable seizure in 38% was recognized by the referring physician in only one case. Unrecognized events included: absence seizure (2), myoclonic (5), and partial complex (8). An EEG obtained in all children with seizures was abnormal in 41%. EEGs obtained early (<48 hours) showed abnormalities in 47% compared to 44% of those obtained late (>48 hours), with no significant difference. (Hamiwka LD, Singh N, Niosi J, Wirrell EC. Diagnostic inaccuracy in children referred with “first seizure”: Role for a First Seizure clinic. **Epilepsia** June 2007;48:1062-1066). (Reprints: Dr Lorie Hamiwka, Alberta Children's Hospital, Division of Pediatric Neurology, 1820 Richmond RD SW, Calgary, AB T2T 5C7, Canada).

COMMENT. Diagnostic inaccuracies in children with “first seizures” are common in general practice, one quarter of patients incorrectly diagnosed as having a seizure rather than a nonepileptic event, while a diagnosis of epilepsy is missed in over one-third. The authors recommend that children with a first seizure should be seen by a neurologist.