mg/kg/day for 33-81 months developed retinal pigmentation, hypopigmented retinal spots, and optic atrophy. Visual evoked potentials, performed in 21, were abnormal initially in 2 and became abnormal during VGB therapy in 14. Electroretinography and electro-oculography were not available. (Koul R, Chacko A, Ganesh A, Bulusu S, Al Riyami K. Vigabatrin associated retinal dysfunction in children with epilepsy. Arch Dis Child Dec 2001;85:469-473). (Respond: Dr R Koul, Department of Child Health (Division of Paediatric Neurology), Sultan Qaboos University Hospital, Al Khod, PO Box 38, 123, Sultanate of Oman).

COMMENT. Regular eye examination including perimetry is recommended in patients treated with vigabatrin. Infants with epileptic syndromes and mental retardation, in whom perimetry is not appropriate, should receive funduscopic examination at least every 3 to 6 months, and retinography, electro-oculography, and VER when available. Retinal changes and visual field constriction associated with VGB can be irreversible, and risks may outweigh benefits if alternative less toxic treatments are available. (see Ped Neur Briefs Dec 2001;15:94-95).

X-LINKED MENTAL RETARDATION AND EPILEPSY SYNDROME

A kindred of 7 affected male infants with an X-linked mental retardation and epilepsy syndrome (XMRE), distinct from X-linked West and other mental retardation-epilepsy syndromes, is reported from the University of Michigan, Ann Arbor, MI. The locus for the new syndrome is identified on chromosome Xp21.1-p11.4. The tetraspanin gene, implicated in nonspecific mental retardation, was not involved. Seizures were primarily generalized, tonic-clonic, and atonic and began at 4 to 14 months of age (average age, 6.8 months). None had infantile spasms or hypsarrhythmia, and other than mental retardation and epilepsy affecting all patients, and mild rigidity and ataxia in 2, the neurologic examination was unremarkable. MRI, EMG, NCS, and laboratory metabolic tests were normal. (Hedera P, Alvarado D, Beydoun A, Fink JK. Novel mental retardation-epilepsy syndrome linked to Xp21.1-p11.4. Ann Neurol January 2002;51:45-50). (Respond: Dr John K Fink, 5214 Cancer Center Geriatrics Center Building, 1500 E Medical Center Drive, Ann Arbor, MI 48109).

COMMENT. X-linked mental retardation is clinically heterogeneous and is reported in 25% of all MR. The authors cite references to 202 XLMR genetically mapped syndromes and 33 identified XLMR genes. Seizures occur with several XLMR disorders, and West syndrome is sometimes transmitted as an X-linked disorder mapped to a locus on Xp21.3-Xp22,1, and distinct from the present novel family. In patients without recognizable metabolic or developmental brain abnormalities, a search for X-linked genetic factors is important in XMRE syndromes.

AUTOSOMAL DOMINANT EPILEPSY SYNDROME LINKED TO 2p11

A newly recognized autosomal dominant epilepsy syndrome with linkage to chromosome 2p11.1-q12.2 is described in an Italian pedigree of 8 patients reported from the Neurosciences Unit, Institute of Child Health and Great Ormond Street Hospital for Children, London, UK. Onset between ages 12 and 50 years, the syndrome is characterized by distal, semi-continuous rhythmic cortical myoclonus, generalized tonic-clonic and complex partial epileptic seizures (ADCME). The majority had only occasional seizures. Seizures were intractable in 3 who developed mild mental retardation. Valproate was most effective, while carbamazepine controlled seizures but exacerbated myoclonus. Interictal EEG abnormalities were generalized and focal frontotemporal. EMG studies of the

myoclonus confirmed that EMG activity was preceded by EEG activity by 8-15 ms, recorded over the contralateral rolandic area, suggesting a cortical origin. (Guerrini R, Bonanni P, Patrignani A et al. Autosomal dominant cortical myoclonus and epilepsy (ADCME) with complex partial and generalized seizures. A newly recognized epilepsy syndrome with linkage to chromosome 2p11.1-q12.2. Brain December 2001;124:2459-2475). (Respond: Professor R Guerrini, Neurosciences Unit, Institute of Child Health and Great Ormond Street Hospital for Children, The Wolfson Center, Mecklenburgh Square, London WC1N 2AP).

COMMENT. This autosomal dominant cortical myoclonus epilepsy syndrome (ADCME) in an Italian pedigree shares some of the characteristics of familial adult myoclonic epilepsy (FAME) described in pedigrees from Japan, except that FAME is linked to chromosome 8q23.3-q24.1, whereas ADCME linkage is to chromosome 2p11.1-q12.2. Clinical characteristics of ADCME include mainly adult onset, non-progressive course, rhythmic myoclonus enhanced during movement, generalized tonic-clonic seizures, and sometimes complex partial seizures.

OUTCOME OF REFRACTORY STATUS EPILEPTICUS

The records of twenty-two children, ages 4.5 months to 18 years, treated for refractory status epilepticus (RSE) between 1992 and 2000, were reviewed retrospectively at Children's Hospital, Boston. Treatment consisted of high-dose anesthetic agents, including pentobarbital, midazolam, propofol, phenobarbital, alone or in combination, for periods ranging from 2 to 146 days (mean 31 days). Outcome correlated with duration of treatment: of 8 treated for <7 days, only 2 died and 4 returned to baseline neurologic state, whereas of 7 requiring treatment of RES for >31 days, 4 died and only 1 returned to baseline. A total mortality of 32% (7 cases) was related to etiology, age, and EEG. The most frequent etiology was presumed or known viral encephalitis affecting 10 patients, of whom 4 died. Of 4 children <3 years of age, none returned to baseline, 3 died, and one had new neurologic deficits after RSE. Of 6 children older than 10 years, 4 returned to baseline, and only one died. Mortality was 25% in 12 children with focal EEG abnormalities at onset of SE, and 40% in 10 with multifocal or generalized EEG abnormalities. No deaths occurred in 5 seizure susceptible children with remote symptomatic seizures not due to an acute provocation; their diagnoses were mental retardation, Rett's syndrome, old intracranial hemorrhage, atypical Rassmussen's encephalitis, and developmental delay. (Sahin M, Menache CC, Holmes GL, Riviello JJ Jr. Outcome of severe refractory status epilepticus in children. Epilepsia Nov 2001;42:1461-1467). (Reprints: Dr M Sahin, Children's Hospital, 300 Longwood Ave, Enders 250, Boston, MA 02115).

COMMENT. Refractory status epilepticus in childhood carries a high mortality (32%) and morbidity that are related to the etiology, age, EEG, and duration of suppressive treatment. Younger patients with presumed encephalitis and multifocal EEGs require prolonged suppressive therapy and the outcome is especially poor.

The findings were similar in a study of 346 adolescent and adult patients with status epilepticus reported from the Netherlands (Scholtes FB et al. Epilepsia 1995;35:1104-1112). A poor outcome was related especially to the underlying cause and status symptomatic of an acute illness, a duration of >4 hours, the occurrence of medical complications, and inadequate therapy. Of 38 patients who died, 44% had received insufficient therapy.

A study in 74 adults with 83 episodes of status epilepticus at the Neurologial Institute, New York, found a mortality of 21% which was correlated with increased