laughter. Epilepsy began with absence attacks at 7 years of age. A prolonged partial status at 10 years was associated with fronto-temporal focal seizure activity and bilateral spikes on the EEG, compatible with partial, secondarily generalized epilepsy. MRI was normal. Control of absence attacks with valproate was incomplete, and trials of oxcarbazepine and clobazam for complex partial seizures were only partially effective. The karyotype was mosaic 46,XX/46,XX,r(20)(p13q13). (Holopainen I et al. Ring chromosome 20 mosaicism in a girl with complex partial seizures. Dev Med Child Neurol Jan 1994;36:70-83). (Respond: Irma Holopainen MD, PhD, Department of Paediatric Neurology, University Hospital of Turku, Kiinamyllynk, 1-3, SF-20500 Turku, Finland).

COMMENT. Chromosome analysis may be indicated in a child with drug refractory idiopathic epilepsy and learning disabilities, despite the absence of dysmorphic features or other congenital anomalies.

Ring chromosome 20 syndrome is characterized by progressive cognitive impairments, behavior disorders and epilepsy resistant to conventional medications. Trials of newer antiepileptic medications with specificity against partial seizures (eg gabapentin) should provide more complete seizure control. (US Gabapentin Study Group No 5. Gabapentin as add-on therapy in refractory partial epilepsy: A double-blind, placebo-controlled, parallel-group study. Neurology Nov 1993;43:2292-2298). Unlike many antiepileptic agents, gabapentin does not affect serum concentrations of AEDs and may be used as concurrent therapy. It had no hematologic, hepatic, pancreatic, or hypersensitivity adverse effects in this 12-week study of 306 patients.

GOLDBERG-SHPRINTZEN SYNDROME & CEREBRAL DYSGENESIS

A 5-year-old girl with Goldberg-Shprintzen syndrome and an abnormal CT scan suggesting neuronal migration defect or brain dysgenesis is reported from the Departments of Pediatrics, Asahikawa Habilitation Center, and Kitami Red Cross Hospital, Japan. Hirschsprung disease was diagnosed at age 4 days, and congenital heart disease with heart failure at 3 months. Clonic convulsions developed at 5 years. Motor development was severely delayed; she sat at 15 months and was unable to stand at 5 years. She had microcephaly, hypertelorism, broad nasal bridge, high-arched palate, thick eyebrows, and an IQ of 25. CT showed frontal and temporal lobe atrophy. The clinical findings in 8 additional patients are summarized from published reports. No chromosomal abnormalities were detected. (Tanaka H et al. Hirschsprung disease, unusual face, mental retardation, epilepsy, and congenital heart disease: Goldberg-Shprintzen syndrome. Pediatr Neurol Nov/Dec 1993;2:479-81). (Respond: Dr Tanaka, Department of Pediatrics, Asahikawa Habilitation Center for Disabled Children, Shunkodai 2-1, Asahikawa 078, Japan).

COMMENT. Waardenburg (congenital deafness, white forelock and depigmented, joined eyebrows, heterochromia iridum, broad nasal bridge) and Smith-Lemli-Opitz (micrognathia, microcephaly, retardation, broad nose and anteverted nostrils, skeletal and urogenital abnormalities) syndromes have similar facial features to those of G-P syndrome and are sometimes complicated by Hirschsprung disease.