Robinow syndrome with variable neurologic features

To the Editor:

We report a father and daughter with Robinow syndrome characterized by macrocephaly, hypertelorism, brachydactyly, short arms, and mild short stature (Fig. 1). The proposita presented to genetics because dysmorphic features were noted during a hospitalization for complications of status epilepticus. She had a history of macrocephaly with white matter atrophy documented at age 7 months, a seizure with residual hemiparesis at 1 year, and mild developmental delay and continued epilepsy with hypoxic encephalopathy because of complications of seizures at 2½ years.

The father and his sister had first presented in childhood for evaluation of possible fetal Dilantin syndrome. Their mother had taken phenobarbital and Dilantin for a seizure disorder throughout her pregnancies. Both children and their mother were noted to have hypertelorism and mild short stature, but no diagnosis was made at the time. Another child in the family had died with hypoplastic left heart syndrome. At reevaluation the diagnosis of Robinow syndrome was made in the proposita and her father. This was based on macrocephaly, facial appearance, and short limbs with supporting evidence of mild Madelung deformity on x-ray films of the proposita and a minor conotruncal anomaly on echocardiography in the father. Photographs from the original clinic visit of the father suggested the same diagnosis in the paternal grandmother and paternal aunt. Of particular interest in this family are the neurologic complications, which are not a usual part of Robinow syndrome.

Up to 20% of patients with Robinow syndrome may have mental retardation,[1,2] and no distinction has been made between dominant and recessive cases with respect to developmental delays. Seizures have not been reported, although developmental brain dysplasia was reported in one patient with Robinow syndrome[3] and communicating hydrocephalus in two others.[4,5] The proposita had mild developmental delay and "white matter atrophy" on magnetic resonance imaging before her seizure onset and hypoxic episode. The father has no history of seizures, but had learning disabilities in childhood now complicated by sequelae of a head injury. The maternal grandmother had "idiopathic" epilepsy with onset in early childhood.

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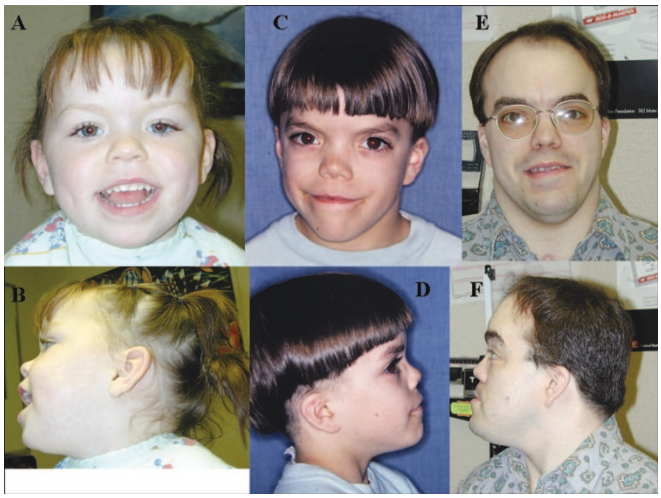

Fig. 1. A and B, Proband age 4 years. C and D, Proband's father age 7.5 years. E and F, Proband's father age 31 years.

Although it is possible that two separate conditions are segregating in this family, it seems likely that these neurologic problems may be part of the variable phenotype of autosomal dominant Robinow syndrome. Caution is recommended in describing the neurologic outlook for infants with Robinow syndrome, and the diagnosis of Robinow syndrome should be considered in patients with suggestive physical characteristics, even if seizures or other unusual neurologic complications are present.

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