COMMENT. Patients with epilepsy and pathogenic structural genomic microarray variants have an objectively more atypical face shape compared with those without. The authors suggest that an evaluation for facial dysmorphism should be part of the clinical work-up for epilepsy. The concept of a “facies epileptica” as defined by Turner (Turner WA. Epilepsy: A Study of the Idiopathic Disease. London: Macmillan and Co; 1907.), is now regarded as unacceptable. Given the heterogeneity of epilepsy, 3D stereophotogrammetry and dense surface models are not expected to identify a specific “face” associated with epilepsy, and actual facial shapes are as varied as the underlying pathogenic structural variants. An objective measure of face shape variation might be used in clinical selection of patients with epilepsy who should be considered for microarray chromosome analysis.

GENETIC NEUROLOGICAL SYNDROMES

EPILEPSY IN MUENKE SYNDROME

Researchers at the National Institutes of Health, Bethesda, MD; Children’s National Medical Center; and George Washington University, Washington, DC present 7 patients with Muenke syndrome complicated by epilepsy. A review of 789 published cases of Muenke syndrome with neurological complications identified epilepsy in 6 cases, with intracranial anomalies in 5. The intracranial anomalies were agenesis of the corpus callosum, hemimegalencephaly, and porencephaly. In the review of 58 patients with Muenke syndrome in the Washington, DC cohort, 7 (12%) had epilepsy and 4 survived neonatal apnea. Patients with Muenke syndrome should be monitored for apnea and seizures. Those with seizures or febrile seizures should undergo neuroimaging, preferably MRI. (Agochukwu NB, Solomon BD, Gropman AL, Muenke M. Epilepsy in Muenke syndrome: FGFR3-related craniosynostosis. Pediatr Neurol 2012 Nov;47(5):355-61). (Respond: Dr Muenke, Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health, Bldg 35, Bethesda, MD 20892. E-mail: mamuenke@mail.nih.gov).