DEVELOPMENTAL DISORDERS

POSTMIGRATIONAL EVOLUTION OF POLYMICROGYRIA

A first in vivo MRI documentation of the postmigrational postnatal evolution of a case of polymicrogyria is reported from the Children's Hospital, Boston, MA. The infant was born premature at 27 weeks gestation. He suffered respiratory distress and cerebral ischemia, and was maintained on a ventilator for 17 days. Ultrasound scans showed bilateral intraventricular hemorrhages at 1 day, and a cystic periventricular leukomalacia that evolved over 3 weeks. MRIs with ultrafine slices were performed at 4 postnatal weeks, term, and at 6 months of age. A minor perisylvian abnormality noted at 4 weeks had developed into extensive bilateral cortical polymicrogyria by 6 months of age, in addition to mineralization of the cystic leukomalacia changes noted earlier. The postnatal evolution of polymicrogyria, associated with the simultaneous occurrence of periventricular leukomalacia, points to an ischemic encephaloclastic mechanism. (Inder TE, Huppi PS, Zientara GP et al. The postmigrational development of polymicrogyria documented by magnetic resonance imaging from 31 weeks' postconceptional age. Ann Neurol June 1999;45:798-801). (Respond: Dr Joseph J Volpe, Neurology Department, Fegan 1103, Children's Hospital, 300 Longwood Ave, Boston, MA 02115).

COMMENT. Congenital bilateral perisylvian polymicrogyria presents with facial diplegia, dysarthria, pseudobulbar palsy, seizures, and retardation. The present case report associated with premature birth, cerebral ischemia, and periventricular leukomalacia is described as unique, although the histopathology is not known.

RM Norman, in Greenfield's Neuropathology (London, Edward Arnold, 1958), refers to sclerotic microgyria (ulegyria) and polygyria among the neuropathological sequelae of birth injury, cerebral ischemia and anoxia. A review of these earlier neuropathological reports is recommended reading.

Genetics of bilateral perisylvian polymicrogyria (BPPMG) is discussed in a report of 6 affected members of 3 consecutive generations of a family. (Borgatti R, Triulzi F, Zucca C et al. Neurology June 1999;52:1910-1913). The severity of the manifestations in one boy affected and the transmission of the disorder through women suggest an X-linked dominant trait. BPPMG is a heterogeneous disorder, in some cases genetically determined, and in others, related to ischemic injury.

MOSAICISM IN TUBEROUS SCLEROSIS COMPLEX

Six families with mosaicism in a series of 62 unrelated families with a mutation in one of the two tuberous sclerosis complex (TSC) genes, TSC1 or TSC2, are reported from the Erasmus University and Hospital, Rotterdam, The Netherlands. In one family the parents showed no clinical signs, but gonadal mosaicism was determined after the diagnosis of TSC was made in 3 children. The exclusion of signs of TSC in the parents reduces the likelihood of a mosaic mutation parental carrier from 10% to 2%. In 5 families with somatic mosaicism, TSC was diagnosed in the parent after the child's case was identified. The genetic counseling implications of mosaicism are discussed. (Verhoef S, Bakker L, Tempelaars AMP et al. High rate of mosaicism in tuberous sclerosis complex. Am J Hum Genet June 1999;64:1632-1637). (Reprints: Dr S Verhoef, MGC Department of Clinical Genetics, Erasmus Academic Hospital, PO Box 1738, 3000 DR Rotterdam, The Netherlands).