

A nonsense mutation of the *ATRX* gene causing mild mental retardation and epilepsy. *Ann Neurol* Jan 2000;47:117-121). (Respond: Dr Gibbons, Nuffield Department of Clinical Biochemistry and Cellular Science, University of Oxford, John Radcliffe Hospital, Headington, Oxford, OX3 9DU, UK).

COMMENT. ATR-X syndrome is clinically heterogeneous. The absence of typical facial dysmorphism does not preclude the diagnosis in a child with mental retardation.

## **BILATERAL FRONTAL POLYMICROGYRIA WITH MENTAL RETARDATION AND EPILEPSY**

Thirteen patients with symmetric polymicrogyria of both frontal lobes are reported from King's College, University of London, UK; University of Pisa, Italy; University of California-San Francisco, CA; UAE University, United Arab Emirates, and University of Chicago, IL. Clinical characteristics included developmental delay, spastic quadriplegia, impaired language development, mental retardation, and epilepsy. Two had consanguineous parents. Head circumference was normal in all but one. Age at examination ranged from 10 months to 32 years. MRI showed irregular infoldings of the cerebral surface with abnormally thick cortex of the frontal lobes bilaterally. EEGs in 5 with epilepsy showed bilateral frontal slowing, sharp waves and spike-and-wave activity. (Guerrini R, Barkovich AJ, Sztriha L, Dobyns WB. Bilateral frontal polymicrogyria; a newly recognized brain malformation syndrome. *Neurology* February (2 of 2) 2000;54:909-913). (Dr Renzo Guerrini, Academic Neuroscience Centre, King's College Hospital, Denmark Hill, London SE 59RS, UK).

COMMENT. Bilateral frontal polymicrogyria is described as a new syndrome, distinct from polymicrogyria involving perisylvian and parasagittal parieto-occipital regions. The majority are detected by MRI in early childhood during investigation for mental and motor retardation or spastic quadriplegia and epilepsy, and some are genetically determined.

## **DOUBLE CORTEX SYNDROME WITH MENTAL RETARDATION AND EPILEPSY**

Magnetic resonance imaging was used to differentiate 30 female sporadic patients with double cortex (DC) syndrome examined at Beth Israel Deaconess Medical Center, Children's Hospital, Boston. Ages ranged from 4 to 46 years. Age at onset of seizures was 6 months to 12 years. Mental retardation ranged from mild to moderate or severe. MRI and genetic tests differentiated patients into four groups: anterior biased/global DC with doublecortin mutation (53%); anterior biased/global DC without mutation (27%); posterior biased DC without mutation (10%); and limited/unilateral DC without mutation (10%). Other genetic loci or mosaicism at the doublecortin locus may be responsible for the heterogeneity of DC syndrome. (Gleeson JG, Luo RF, Grant PE et al. Genetic and neuroradiological heterogeneity of double cortex syndrome. *Ann Neurol* February 2000;47:265-269). (Respond: Dr CA Walsh, Division of Neurogenetics, Beth Israel Deaconess Medical Center/Harvard Medical School, 77 Avenue Louis Pasteur, Boston, MA 02115).

COMMENT. Double cortex syndrome can represent a genetically heterogeneous group of mental retardation syndromes, 50 per cent showing an identifiable DCX mutation. MRI scans in patients with mutation have an anterior biased subcortical band and overlying pachygyria, whereas some without