NEUROCUTANEOUS SYNDROMES

CORTICAL MALFORMATIONS IN NEUROFIBROMATOSIS TYPE 1

Different types of malformations of cortical development in three cases of neurofibromatosis 1 (NF1) are reported from the University of Siena, Italy. Patient 1, a 3-year-old boy with infantile spasms, café-au-lait spots, Lisch nodules, and skin fibromas had an abnormal MRI with a large dysplasia involving the right temporoinsuloparieto-occipital cortex. Patient 2, a 19-year-old woman with a history of infantile spasms, café-au-lait spots, Lisch nodules, axillary and inguinal freckles, and neurofibromas had an MRI showing a periventricular band of heterotopic gray matter and overlying pachygyric cerebral cortex. Patient 3, an 18-year-old man with mental retardation, infantile febrile seizures, left partial clonic and generalized seizures at age 8 years, EEG paroxysmal activity on the left temporal area, had an MRI showing a left perisylvian polymicrogyria. Neurofibromin may play a role during several stages of cortical development. (Balestri P, Vivarelli R, Grosso S et al. Malformations of cortical development in neurofibromatosis type 1. **Neurology** December (2 of 2) 2003;61:1799-1801). (Reprints: Dr Paolo Balestri, Department of Clinical Pediatrics, University of Siena, Viale M Bracci, Le Scotte, 53100 Siena, Italy).

COMMENT. Severe cortical malformations are considered rare in NF1. When present they can be extremely variable and complex, and are associated with drug resistant epilepsy and mental retardation. According to the Barkovich classification, cited by the authors (Barkovich AJ et al. **Neurology** 2001;57:2168-2178), the first patient reported above had a malformation showing abnormal neuronal and glial proliferation or apoptosis. The abnormality in the second patient could be related to abnormal neuronal migration, and the third is an example of an anomaly caused by defective cortical organization. These malformations represent different stages of cortical development, all resulting from deficiencies of neurofibromin.

Malformations of cortical development (MCD) in epilepsy are reviewed from the Institute of Neurology, Queen Square, London (Sisodiya SM. **Lancet Neurology** January 2004;3:29-38). The most common MCDs include focal cortical dysplasias, periventricular heterotopia, polymicrogyria, band heterotopia, lissencephaly, dysembryoplastic neuroepithelial tumors, and microdysgenesis. Subtle MCD not detectable on MRI and identified in surgical tissue samples may sometimes underlie epilepsies termed cryptogenic.

COGNITIVE AND FINE MOTOR DEFICITS AND MRI HYPERINTENSITIES IN NEUROFIBROMATOSIS TYPE 1

The relationship between cognitive impairment, fine motor deficits, and T2-weighted MRI intensities in neurofibromatosis type 1 (NF1) was investigated in 100 patients and 100 healthy controls in a study at University Hospital of Munster, Germany. T2 hyperintensities (T2H) were found in 66% of 56 patients younger than 16 and in 48% of 44 patients older than 16. As a group, patients with NF1 had normal scores on the WISC-R and WAIS-R. Patients with normal MRI had close to the mean IQ of the normal population whereas those