GENETIC/ENVIRONMENTAL INFLUENCES IN FRAGILE X DISEASE

The genetic and environmental factors influencing cognitive outcomes in 120 children (80 boys and 40 girls) with the fragile X full mutation and their unaffected siblings were determined by in-home evaluations and reported from the Departments of Psychiatry and Pediatrics, Stanford University, CA. The MPIQ (mean full-scale IQ of biological parents) was the primary determinant of the IQ of comparison siblings, and the quality of the home environment had an additional but small effect on verbal development. Using multiple regression analyses, cognitive outcome of girls with fragile X was correlated with the mean IQ of their parents, and to a lesser degree with the quality of home environment. FMR 1 protein % (FMRP) was correlated with girls' levels of distractibility. Girls with fragile X had higher IQs than boys, with relatively stronger Verbal Scales. The affected boys' Performance IQ was related to the mean parental IQ, while the boys' Full Scale IQ was correlated with FMRP %. The quality of boys' home environments affected their cognitive outcomes more so than in affected girls. (Dyer-Friedman J, Glaser B, Hessl D et al. Genetic and environmental influences on the cognitive outcomes of children with fragile X syndrome. J Am Acad Child Adolesc Psychiatry March 2002;41:237-244). (Respond: Dr Dyer-Friedman, Department of Psychiatry, Child Division, 401 Quarry Road, Stanford University School of Medicine, Stanford, CA 94305).

COMMENT. Both genetic/parental and environmental factors are significant in the prediction of cognitive outcome of children with fragile X syndrome. The specific factors influencing IQ are different in girls and boys.

Brain anatomy in fragile X syndrome. MRI scans and cognitive testing were performed in 37 children and adolescents with fragile X syndrome at Stanford University. Decreases in grey matter and increases in white matter were age- and gender-related. Caudate and ventricular CSF volumes were significantly enlarged, and caudate volumes decreased with age. IQ scores and volumes of cortical and subcortical grey matter were not significantly correlated, but were different from the correlations observed in normal children. (Eliez S, et al. Brain 2001;124:1610-1618).

PERINATAL DISORDERS

SEIZURE-ASSOCIATED BRAIN INJURY IN PERINATAL ASPHYXIA

Brain injury in term infants with neonatal asphyxia and seizures was evaluated by MRI and MRS at the University of California, San Francisco. Seizure severity was scored by seizure frequency and duration, EEG, and anticonvulsant therapy. Impairment of cerebral metabolism and neuronal integrity in the intervascular boundary zone and basal nuclei were measured by lactate/choline and N-acetyl aspartate/choline. Clinical seizures occurred in 37% of 90 infants studied. At 6 days of age (range, 1-13 days), seizure severity was associated with increased lactate/choline in both brain locations tested, and a diminished N-acetylaspartate/choline in the intervascular boundary zone but not in basal ganglia. The severity of seizures was independently associated with chemical evidence of brain injury and was not limited to the structural brain damage detected by MRI. (Miller SP, Weiss J, Barnwell A et al. Seizure-associated brain injury in term newborns with perinatal asphyxia. Neurology February (2 of 2) 2002;58:542-548). (Reprints: Dr AJ Barkovich, Department of Radiology, University of