HEREDODEGENERATIVE DISEASES

GENETICS OF X-LINKED ADRENOLEUKODYSTROPHY

An 8.5-year-old girl with cerebral X-linked adrenoleukodystrophy is reported from Ben-Gurion University, Beer Sheva, Israel, and Johns Hopkins University, Baltimore, MD. Early development was normal, she could read by 6 years, but severe behavioral problems and deterioration of academic performance developed by 8 years of age. The symptoms of "school phobia" included separation anxiety, oversensitivity to noise, and isolation from peers and teachers. Neurologic exam was normal and intelligence scores by WISC-R were low normal. Achievement was impaired by distractibility and visuospatial defects. Cortisol response to IV ACTH was normal, ruling out adrenal insufficiency. MRI revealed diffuse white matter involvement, most prominent in frontal regions. Very long chain fatty acids in plasma and skin fibroblasts were elevated. Cytogenetic analyses on peripheral lymphocytes showed a deletion at Xq27.2-ter and the unexpected demonstration of a de novo deletion in her paternal X chromosome, involving all of Xq28 and part of Xq27. In combination with the abnormality on the maternal X chromosome, this caused failure of expression of functional ALDP, similar to that in affected males. Neurological and psychological status was stable at 18 months after bone marrow transplant. (Hershkovitz E, Narkis G, Shorer Z et al. Cerebral X-linked adrenoleukodystrophy in a girl with Xq27-ter deletion. Ann Neurol August 2002;52:234-237). (Respond: Eli Hershkovitz MD. Pediatric Department, Soroka University Medical Centre, Beer Sheva, Israel).

COMMENT. Cytogenetic studies are recommended in severely symptomatic X-linked adrenoleukodystrophy heterozygotes. Treatment with bone marrow transplant as recommended in boys with ALD can be beneficial in affected girls.

SEIZURE DISORDERS

IRON INSUFFICIENCY AND FEBRILE SEIZURES

The significance of iron status as a possible risk factor for a first febrile seizure (FFS) was investigated at Jordan University and King Hussain Medical Center, Irbid, Jordan. Mean ferritin level in this prospective study of 75 children with FFS (29,5 +/- 21.3 mcg/L) and 75 matched controls (53.3 +/- 37.6 mcg/L) was significantly decreased (P=0.0001). A plasma ferritin level of <30 mcg/L was significantly more prevalent among children with FFS (49 of 75) than controls (24 of 75). Mean levels of Hgb, MCV, and MCH were lower in FFS cases, and a higher proportion of FFS cases had an Hgb <110 g/L, MCV <72 fL, and MCH <24 pg, but differences were not significant. The findings suggest a possible role for iron insufficiency in FFS. (Daoud AS, Batieha A, Abu-Ekteish F et al. Iron status: A possible risk factor for the first febrile seizure. Epilepsia July 2002;43:740-743). Dr A Daoud, Department of Pediatrics, Jordan University of Science and Technology, Irbid, Jordan).

COMMENT. Plasma ferritin is used as a reliable measure of iron deficiency and total body iron status. Decreased plasma ferritin levels in children with a first febrile seizure are not explained by fever, since the mean peak temperature on admission was similar in patients and controls. Other neurologic disorders in which iron-deficiency anemia may play a role include breath-holding spells, developmental delay, and behavior and attention disorders.