
COMMENT. In an editorial in the same issue Peterson HDA and Funkhouser JD of the University of South Alabama, Mobile, AL refer to their previously published proposal that essentially all clinical manifestations of ataxia-telangiectasia, including the degeneration of the CNS, are a consequence of a defect in genetic recombination. (Immunol Today 1989; 10:313-5). Elucidation of the molecular abnormalities of the lymphocytes in patients with ataxia-telangiectasia may reveal molecular mechanisms responsible for the cellular differentiation of lymphocytes and other cell systems. The gene responsible for ataxia-telangiectasia has been localized to chromosome 11q22-23. These studies at the molecular level bring new insight in lymphocyte differentiation and the immune disorder that characterizes ataxia-telangiectasia. Absence of tonsils in a child with ataxia should prompt the determination of immunoglobulins.

METACHROMATIC LEUKODYSTROPHY

A ten year old girl with metachromatic leukodystrophy in whom neurophysiologic function and sulfatide metabolism had improved after she received a bone marrow transplant five years before is reported from the Bone Marrow Transplantation Program, Department of Pediatrics and Division of Pediatric Neurology, University of Minnesota, MN and other centers. The diagnosis was confirmed by enzyme analysis at eight months of age after an older sister had been found to have the disease. Serial MRI of the head obtained before and after bone marrow transplantation showed no further deterioration of white matter. Sural nerve specimens obtained by biopsy before and two years after transplantation showed less accumulation of lipid in the macrophages on electron microscopy. Sulfatide levels in the CSF were within normal limits at seven and ten years of age. Asymptomatic infants, children and young adolescents who are found to have the disease after it has been diagnosed in an older sibling should be considered for bone marrow transplantation. (Krivit W et al. Treatment of late infantile metachromatic leukodystrophy by bone marrow transplantation. N Engl J Med Jan 4, 1990; 322:28-32).