AICARDI-GOUTIERES SYNDROME

The clinical, radiological, and biological features of Aicardi-Goutieres syndrome in 27 patients are reviewed from the Neuropediatric Unit, Hopital des Enfants Malades, Paris, France. The onset was within the first 4 months of life in 19. The head circumference was normal at birth, but 21 developed microcephaly during the first year. CTs showed severe, progressive brain atrophy in all patients, and variable calcification of the basal ganglia. CSF lymphocytosis was chronic and persisted beyond 1 year in 7 patients. High levels of interferon-a occurred in serum and CSF in 14. Nineteen patients who survived, 6 older than 10 years, are severely disabled. Neuropathological findings in 2 patients showed foci of necrosis and diffuse demyelination, without inflammation. An autosomal recessive inheritance is suspected. (Goutieres F, Aicardi J, Barth PG, Lebon P. Aicardi-Goutieres syndrome: an update and results of interferon-a studies. Ann Neurol Dec 1998;44:900-907). (Respond: Dr Goutieres, Neuropediatric Unit, Hopital des Enfants Malades, 149 Rue de Sevres, 75743 Paris Cedex 15, France).

PHENOTYPES OF JUVENILE BATTEN DISEASE

The phenotypes of 10 Finnish juvenile neuronal ceroid lipofuscinosis (JNCL; late-onset Batten disease) patients were correlated with the genotypes in a study at Helsinki University, Finland; and the Rayne Institute, University College, London, UK. JNCL is manifested as three phenotypes: classic, delayed classic, and protracted JNCL, with mainly ocular symptoms and slower mental and motor decline. All are compound heterozygotes for 5 rare mutations and the major 1.02-kb deletion in the CLN3 gene. A novel deletion of exons 10 through 13 was present in 6 patients in 3 families, all having a similar clinical course. The development of blindness showed the greatest familial heterogeneity, from 6 to 15 years. (Lauronen L, Munroe PB, Jarvela I et al. Delayed classic and protracted phenotypes of compound heterozygous juvenile neuronal ceroid lipofuscinosis. Neurology Jan 1999;52:360-365). (Reprints: Dr Leena Lauronen, BioMag Laboratory, PO Box 508, Fin-00029, HYKS, Finland).

COMMENT. Late-onset Batten disease (JNCL) is an autosomal recessive, progressive encephalopathy of childhood, with ceroid and lipofuscinlike material in neural and nonneural tissues. The gene is on chromosome 16p11.2-12.1.