ALAd-Deficiency Porphyria (ADP)

OVERVIEW & SYMPTOMS

ADP is the least common of all the porphyrias with fewer than 10 cases documented to date. This is an autosomal recessive disease, whereas the other three acute porphyrias are autosomal dominant. All of the reported cases have been males, in contrast to the other AHPs.

A severe deficiency of the enzyme δ-aminolevulinic acid dehydratase (ALAD) causes an increase of 5-aminolevulinic acid (ALA) in the liver, other tissues, blood plasma, and urine. In addition, urine coproporphyrin and erythrocyte protoporphyrin are increased.

TREATMENT & MANAGEMENT

Treatment is the same as in the other AHPs. Liver transplantation alone has not been of great benefit in the one patient with ADP so treated, but a recent report from Holland indicated that IV hemin and hypertransfusions and hydroxycarbamide, the latter to decrease bone marrow overproduction of porphyrin precursors, was effective in another boy with ADP.

FAMILY TESTING & COUNSELING

ADP is the only AHP that is autosomal recessive. All children of an affected individual will be carriers of ADP, but are unlikely to develop any symptoms.