Letter to the Editor: Frequency of the PD Allele

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In our report on the prenatal diagnosis in a family with pseudo arylsulfatase A deficiency at risk for metachromatic leukodystrophy (MLD), we indicated that it was our impression that the frequency of the PD allele might be equal to or higher than the frequency of the MLD allele (1). The observations of Zlotogora *et al.* and our recent involvement in five additional such prenatal diagnosis [one a repeat pregnancy in the index case (1)] have reinforced our view. A systematic study of the nuances of the present allelism requires identification of individuals with different genotypes. However, existing enzyme assay techniques do not allow differentiation between the MLD heterozygote and the PD heterozygote, and unequivocal differentiation between the PD homozygote and the MLD-PD mixed heterozygote is problematical. The approach taken by Zlotogora *et al.*, namely studies of

parents of known pseudodeficient subjects, will allow the use of genetic considerations as well as biochemical information to identify individuals with different genotypes. In one family, genotype prediction was possible by genetic criteria alone. The positive identification of individuals with the various PD genotypes will greatly facilitate development of biochemical protocols applicable for population studies.

REFERENCES AND NOTES

1. Kihara, H., Ho, C. K., Fluharty, A. L., Tsay, K. K., and Hartlage, P. L.: Prenatal diagnosis of metachromatic leukodystrophy in a family with pseudo arylsulfatase A deficiency by the cerebroside sulfate loading test. *Pediatr. Res.*, 224 (1980).

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