Identification of an Apa I polymorphism within the human adenosine deaminase (ADA) gene

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T.Gribbin, E.Chottiner, D.Ginsburg¹ and B.Mitchell

Department of Internal Medicine and ¹The Howard Hughes Medical Institute, University of Michigan Medical Center, Ann Arbor, MI 48109, USA

SOURCE/DESCRIPTION: The probe used is a 1.4 kb adenosine deaminase (ADA) cDNA probe cloned into the Hind III site of pBr322 (1). This RFLP has been identified in human genomic DNA isolated from peripheral blood mononuclear cells and cultured B lymphoblasts.

POLYMORPHISM: Apa I identifies a two allele polymorphism within the fourth intron of the ADA gene. Southern blot analyses demonstrate 3.7 kb and 2.9 kb bands (A1) or a single 6.6 kb band (A2). Southern blots of heterozygote DNA demonstrate both A1 and A2.

FREQUENCY: Allele frequencies as estimated from the analysis of 20 unrelated individuals are 0.8 (A1) and 0.2 (A2), respectively.

CHROMOSOMAL LOCALIZATION: The human ADA gene has been assigned to the long arm of chromosome 20 (2).

MENDELIAN INHERITANCE: Four of seven members of a family with hereditary overproduction of erythrocyte ADA were heterozygous for the Apa I RFLP, and a co-dominant segregation pattern was observed (3).

PROBE AVAILABILITY: The ADA cDNA probe is available through the American Type Culture Collection.

REFERENCES: 1) Daddona, PE *et al.* (1984), *J. Biol. Chem.* 259, 12101-12106; 2) Tischfield, JA *et al.* (1974), *Hum Hered.* 24, 1-11; Chottiner, EG *et al.* (1989), *The Red Cell*, G. Brewer, ed., in press.

ACKNOWLEDGEMENTS: This work is supported by grant 1-RO1-AI24012 from the National Institutes of Health.