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## A Pvu II RFLP in the human ADH3 gene

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**SOURCE/DESCRIPTION:** A 1.4 Kb fragment containing the third exon of the ADH3 gene was cloned into pUC8 to give plasmid pADH74. The insert can be excised by Eco RI/Bam HI digestion.

**POLYMORPHISM:** Pvu II identifies 2 allele polymorphisms (A1: 5.4 Kb, A2: 4.3 Kb). 5.4 allele is superimposed on a constant band derived from the third exon of ADH2. Given the high level of homology between the ADH2 and ADH 3 genes, a faint 5.4 Kb band is visible also in 4.3 Kb homozygotes.

**FREQUENCY:** Estimated on 24 unrelated Caucasians:  
 Pvu II A1 allele (5.4 Kb): 0.56  
 Pvu II A2 allele (4.3 Kb): 0.44

**NOT POLYMORPHIC FOR:**  
 EcoRI MspI AluI Sau3a

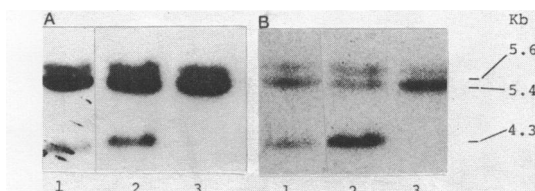
**CHROMOSOMAL LOCALISATION:**  
 Localized on chromosome 4q21-q23

**MENDELIAN INHERITANCE:**  
 Co-dominant segregation shown in 5 informative families

**PROBE AVAILABILITY:**  
 Available from ATCC (57220)

**OTHER COMMENTS:** Linkage disequilibrium is present with the Xba I polymorphism detected by pADH74 ( $\chi^2$ : $P < 0.02$ ). 5.4 Kb Pvu II allele is associated with 4.4 Kb Xba I allele, 4.3 Kb Pvu II allele with 3.3 Kb Xba I allele.

**REFERENCE:**  
 -Murray J.C., Shiang R, Carlock L.R., Smith, M, Buetow K.H., Hum. Genet. 76:274-277 (1987).



**FIGURE LEGEND:** Autoradiography of Southern blots of Pvu II digested genomic DNA from three individuals, probed with pADH74 (panel B) or with a probe for exon 3 of ADH2 (panel A). In both panels lane 1 is a 5.4/4.3 Kb Pvu II heterozygote, lane 2 is 4.3 Kb homozygote and lane 3 is a 5.4 Kb homozygote. The faint 5.6 Kb band visible in all lanes in both panels is likely to be a homologous fragment from exon 3 of ADH1.