TaqI polymorphism in the human LDL receptor gene

Kimiko Yamakawa, Takaaki Okafuji, Yukio Iwamura, David W. Russell¹ and Hideo Hamaguchi

Department of Human Genetics, Institute of Basic Medical Sciences, University of Tsukuba, Niihari-gun, Ibaraki-ken 305, Japan and ¹Department of Molecular Genetics, University of Texas Health Science Center, 5323 Harry Hines Blvd, Dallas, TX 75235, USA

SOURCE AND DESCRIPTION : The probe that contains exon 1 to 8 was generated from pLDLR3. pLDLR3 contains a full-length LDL receptor cDNA (Yamamoto *et al.*, 1984)

POLYMORPHISM : *TaqI* identifies a bi-allelic polymorphism with a band of 1.7kb and 0.6kb.

FREQUENCY : Studies in 55 unrelated normal Japanese.
1.7kb fragment 0.69
0.6kb fragment 0.31

NOT POLYMORPHISMS : In 50 individuals no polymorphisms were detected with *Bam*HI, *Bgl*II, *Eco*RI, *Eco*RV and *Msp*I.

CHROMOSOMAL LOCALIZATION : 19q13.1-13.3 (Lindgren *et al.*, 1985).

MENDELIAN INHERITANCE : Co-dominant segregation observed in 4 families (19 individuals).

COMMENTS : In fifty people studied, this *TaqI*-RFLP is in linkage equilibrium with the previously reported four RFLPs (*Pvu*II, *Pst*I, *Ava*II, *Nco*I).

REFERENCES : (1). Yamamoto, T., *et al.* (1984) *Cell* 39 : 27-38.
(2). Lindgen, V., *et al.* (1985) *PNAS* 82 : 8567-8571.

Acknowledgement : Supported by the Grant-in-Aid for Scientific Research (No. 61571088) from the Ministry of Education, Science and Culture of Japan.