

AvaII RFLP at the human apolipoprotein CII (APO CII) gene locusR.G.Korneluk, H.L.MacLeod, S.C.Leblood, N.L.Monteith, F.E.Baralle¹ and A.G.W.HunterGenetics Division, Children's Hospital of Eastern Ontario, Ottawa, Ontario, Canada and ¹Sir William Dunn School of Pathology, University of Oxford, South Parks Road, Oxford OX1 3RE, UK

SOURCE AND DESCRIPTION OF CLONE: Human apolipoprotein CII (apo CII) cDNA, 489 bp, isolated from a human liver cDNA library and subcloned into the PvuII site of pAT153/PvuII/8. (1)

POLYMORPHISM: AvaII identifies a simple two-allele polymorphism, with bands at 0.6 kb and 0.4 kb. Two invariant bands are also detected at 1.4 kb and 0.25 kb. (see Figure)

FREQUENCY: Studied in 83 unrelated Caucasians:
 0.6 kb allele 0.55
 0.4 kb allele 0.45

NOT POLYMORPHIC FOR: ApaI, AvaI, BclI, BcnI, BglI, BglII, BstNI, DraI, EcoRI, HinfI, KpnI, MboI, MspI, PstI, RsaI, ScaI, ScrFI, StuI, XbaI, XmnI, studied in 10 unrelated Caucasians.

CHROMOSOMAL LOCALIZATION: 19cen - 19q13.2 (HGM8).

MENDELIAN INHERITANCE: Codominant segregation has been shown in 23 Myotonic Dystrophy families, totalling 163 individuals.

PROBE AVAILABILITY: Requests for this probe should be directed to F.E. Baralle, at the above address.

OTHER COMMENTS: Stringency of post-hybridization washes: 0.2xSSC, 0.1% SDS, 50°C.

REFERENCE: (1) Sharpe, C.R. et.al. (1984). Nucl. Acids Res. 12(9): 3917-3932.

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