DNA polymorphic sites in the human ApoAI-CIII-AIV cluster: Taq I and Ava I

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SOURCE AND DESCRIPTION OF CLONE: The probe was a 2.2. Pst I genomic fragment containing human ApoAI sequences cloned in plasmid PSV2-GPT (Karathanasis et al., 1983).

POLYMORPHISM: 1. Taq I digestion of genomic DNA and hybridisation with the ApoAI probe detects a two allele polymorphism with allelic fragments of 4.8 and 8.6 kb. The frequency of the polymorphic site in 44 unrelated Caucasians was:

Taq I site present (4.8 kb fragment) : 0.72
Taq I site absent (8.6 kb fragment) : 0.68

2. Ava I digestion of genomic DNA and hybridisation with the ApoAI probe detects a two allele polymorphism with allelic fragments of 6 and 15 kb. The frequency of the polymorphic site in 17 unrelated Caucasians was:

Ava I site present (6 kb fragment) : 0.32
Ava I site absent (15 kb fragment) : 0.68

CHROMOSOMAL LOCALISATION: The ApoAI-CIII-AIV cluster has been localised to the long arm of human chromosome 11 (Cheung et al., 1984).

MENDELIAN INHERITANCE: Co-dominant segregation demonstrated in more than 10 Caucasian families.

PROBE AVAILABILITY: Request for probe to S.K.K. at the above address.

REFERENCE : Cheung P, Kao FT, Law ML, et al. Proc. Natl. Acad. Sci. USA. 81:508-511, 1984.
Karathanasis SK, Zannis VI, Breslow JL. Proc. Natl. Aca. Sci. USA. 80:6147-6151, 1983.

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