Two new ApoB gene polymorphisms: Rs1 and Rs2

J.J.Genest Jr, M.J.Ordovas, A.H.Robbins, D.C.King, P.M.Frossard and E.J.Schaefer

Lipid Metabolism Laboratory, Tufts University, Boston and California Biotechnology, Mountain View, CA, USA

SOURCE/DESCRIPTION : 2.0 kb fragment intestinal cDNA clone I(2)1 in pUC18. (1)

POLYMORPHISM : RsaI (BRL, Bethesda Md) digests identifies 7 fragments: 2.2, 2.0, 1.4, 1.2, 0.7, 0.6 and 0.4 kb, spanning a 12 kb region of ApoB. Rs1 detected at 2.7 kb (fig A), Rs2 at 1.8 kb (fig B).

FREQUENCY : Rs1: 2/67 unrelated caucasians.
Rs2: 0/31 unrelated caucasians with angiographically normal coronary arteries, 6/36 with coronary artery disease.

NOT POLYMORPHIC FOR:

CHROMOSOMAL LOCALISATION : 2p23-24 by in-situ hybridization (2)

MENDELIAN INHERITANCE : Co-dominant segregation observed for Rs1 and Rs2

PROBE AVAILABILITY: Protter A.A.

OTHER COMMENTS : Rs1 localizes to near exon 26 with 1.3 kb 5' fragment of SstI digest of I(2)1. Rs2 localizes to near exon 18 with 0.2 kb 3' fragment of SstI digest of I(2)1.

REFERENCE : (1) Protter A.A et al. PNAS 83(1986)5678-5682
(2) Law S.W. et al. PNAS 82(1985)8340-8344

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