Two Pstl RFLPs in the PCCB gene on the long arm of chromosome 3

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SOURCE/DESCRIPTION: The probe is a ~1.2 kb BamHI fragment from pPCC41A2, a cDNA clone encoding the beta polypeptide of propionyl-CoA carboxylase (1). An EcoRI RFLP in PCCB has been described previously (1).

POLYMORPHISM: PstI (CTGCAG) detects two polymorphisms which are in equilibrium. One two-allele polymorphism has bands at 10.2 kb (B1) or 7.8 kb (B2). A second two-allele polymorphism has bands at 6.0 kb (C1) or 1.7 kb (C2). This second RFLP is in strong disequilibrium with the previously described EcoRI polymorphism (1). Invariant bands are detected at 12.0 kb, 8.1 kb, 4.2 kb, and 3.0 kb.

FREQUENCIES: 130 chromosomes were scored from 65 unrelated Caucasian individuals.

10.2 kb allele (B1): 0.90 7.8 kb allele (B2): 0.10

6.0 kb allele (C1): 0.47 1.7 kb allele (C2): 0.53

NOT POLYMORPHIC FOR: In a screen of six unrelated individuals, PCCB was not polymorphic for XbaI, DraI, BglII, and RsaI.

CHROMOSOMAL LOCALIZATION: PCCB has been assigned to chromosome $3q13.3 \longrightarrow q22 (1,2)$.

MENDELIAN INHERITANCE: Co-dominant inheritance demonstrated in several large, three-generation families (C.E.P.H.).

PROBE AVAILABILITY: Available from the ATCC probe repository.

OTHER COMMENTS: Low background with final wash at 65°C in 0.1 x SSC/0.1% SDS.

REFERENCES: (1) Lamhonwah et al., Proc. Natl. Acad. Sci. USA 83:4864-4868 (1986).

(2) Kraus et al., Proc. Natl. Acad. Sci. USA 83: 2047-2051 (1986).

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