Human parathyroid hormone-like peptide gene: HindIII and TaqI RFLPs

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Source/Description: sH-4, a 2 kb HindIII-HindIII fragment encoding exons V and VI of the human parathyroid hormone-like peptide (hPLP) gene subcloned in pGEM-1 (1).

Polymorphisms: HindIII identifies a multiple allele polymorphism with at least 5 bands of sizes 2.3 kb (allele 1), 2.25 kb (allele 2), 2.2 kb (allele 3), 2.15 kb (allele 4) and 2.10 kb (allele 5). TaqI identifies the same polymorphism with bands of sizes 1.65 kb (allele 1), 1.6 kb (allele 2), 1.55 kb (allele 3), 1.5 kb (allele 4) and 1.45 kb (allele 5).

Frequency: The following frequency of the HindIII polymorphism was estimated from a study of 35 unrelated North Americans of mixed ethnic origin. Allele 1, 12.9%; allele 2, 18.6%; allele 3, 18.6%; allele 4, 12.9%; allele 5, 37.1%: 80% heterozygosity.

Not Polymorphic For: EcoRI, PstI, BclI (tested with a panel of 9 unrelated individuals).

Chromosomal Localization: The hPLP gene has been localized to chromosome 12p12.1-p11.2 by *in situ* hybridization (2, 3).

Mendelian Inheritance: Co-dominant autosomal segregation of the HindIII polymorphic alleles was observed in 3 informative families (14 individuals).

Probe Availability: Contact G.N.Hendy.

References: (1) Yasuda, T. *et al.* (1989) *J. Biol. Chem.* **264**, 7720–7725. (2) Mangin, M. *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 597–601. (3) Suva, L.J. *et al.* (1989) *Gene* **77**, 95–105.

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Southern blot analysis of DNA from 9 unrelated individuals probed with sH4: a) HindIII restriction digest, b) TaqI restriction digest. The alleles are numbered 1–5.

RFLP for intron E of factor XI gene

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Source/Description: Intron E is a 1.4 kb fragment of the Factor XI gene (1). This fragment was synthesized by the polymerase chain reaction (2) with TaqI polymerase after 30 cycles of DNA amplification at an annealing temperature of 60°C with the use of 3' and 5' flanking sequences of intron E. These sequences were: TGGTTGGTGTCCCTGTTTGGGTGTG at the 3' end and AGGCAGTTTCCCAGCCTGGAGCAT at the 5' end (1).

Polymorphism: Hha I identifies a two allele polymorphism with bands at 0.15 kb (A1) and 0.14 kb (A2) and invariant bands at 0.61, 0.46 and 0.32 kb.

Allele Frequencies: Estimated on 30 unrelated Caucasians.

A1 allele (0.15 kb) : 0.40

A2 allele (0.14 kb) : 0.60

Observed PIC Value: 0.37.

Not Polymorphic For: Hph I, Alu I, Hae III, Rsa I.

Chromosomal Localization: The factor XI gene is located at 4q35 (3).

Mendelian Inheritance: Codominant segregation was observed in three families.

Probe Availability: Contact M.G.Butler.

References: (1) Asakai, R. *et al.* (1987) *Biochemistry* **26**, 7221–7228. (2) Saiki, R. *et al.* (1988) *Science* **239**, 487–491. (3) Kato, A. *et al.* (1989) *Cytogenetics and Cell Genetics* **52**, 1–2.

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