TaqI polymorphism at the human complement component C9 gene

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Source and Description of Clone: pHLC9.63, a 2.38 kb C9-cDNA in PAT153/PvuII/8 (1).

Polymorphism: TaqI detects a two allele polymorphism:

A1 = 6.5

A2 = 8.0 + 6.0 kb

Frequency: Estimated from 30 Spanish individuals.

A1 = 0.74

A2 = 0.26

Not Polymorphic For: EcoRI, HindIII, PstI and BamHI.

Chromosomal Localization: 5p (2).

Mendelian Inheritance: Codominant in three 2 generation families.

Probe Availability: American Type Culture Collection (ATCC 59114).

References: 1) DiScipio, R.G. *et al.* (1984) *Biochemistry* **81**, 7298. 2) Abbott, C. *et al.* (1989) *Genomics* **4**, 606.

A BamHI polymorphism at the DBH locus

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Source/Description: DBH A4 is a 2.3 kb dopamine- β -hydroxylase cDNA (1) cloned into the EcoRI site of pUC18.

Polymorphism: BamHI identifies a simple two allele polymorphism, with a variable band at 4.5 kb or 2.0 kb. Constant bands are observed at 7.9 kb, 5.5 kb and 3.0 kb (see figure).

Frequency: Studied in 57 unrelated individuals.

4.5 kb allele = 0.18

2.0 kb allele = 0.82

Not Polymorphic For: HindIII, PstI, EcoRI, PvuII. See also ref. 3.

Chromosomal Localization: Localized to 9q34 by *in situ* hybridization (2).

Mendelian Inheritance: Codominant in 34 meioses (in tuberous sclerosis families).

Probe Availability: Contact Dr J.Mallet, Laboratoire de Neurobiologie Cellulaire et Moléculaire, CNRS, 91198, Gif-sur-Yvette, France.

Other Comments: Two *TaqI* polymorphisms have also been identified with a DBH cDNA probe (3).

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References: 1) Lamouroux, A. *et al.* (1987) *EMBO J.* **6**, 3931-3937. 2) Craig, S.P. *et al.* (1988) *Cytogenet. Cell Genet.* **48**, 48-51. 3) Amato, D. *et al.* (1989) *Nucl. Acids Res.* **17**, 5871.