A new *Ava*II RFLP in the human α -globin gene cluster

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Source/Description: A 1.5 kb *Pst*I fragment isolated from the plasmid p α BR containing the α 1 globin gene was used as hybridization probe (1).

Polymorphism: Restriction analysis of human DNA with the enzyme *Ava*II revealed a polymorphic site located 1 kb upstream from the α 1 globin gene. The presence or absence of this restriction site results in a 0.9 kb or 1.95 kb fragment respectively (see figure: lane 1 -/-; lane 2 +/-; lane 3 +/+).

Frequency: Determined for a total of 25 individuals of Caucasian origin. Allele 0.9 kb: 0.75. Allele 1.95 kb: 0.25.

Chromosomal Localization: 16p13.3 (2).

Mendelian Inheritance: Mendelian inheritance was observed in a total of 6 nuclear families (12 meioses).

Probe Availability: Contact R.Fodde.

Comments: This RFLP may be included with other common polymorphisms for α -globin haplotyping.

Due to the large degree of homology between the α 1 and α 2 globin genes, the possibility exists that a corresponding *Ava*II site located 5' to the α 2 gene might also be present but escape detection due to failure to evaluate differences in the relative intensity of the 0.9 kb fragment. The more common absence of this site results in a 2.4 kb fragment always observed during this study (see Figure).

References: 1) Lauer, J. *et al.* (1980) *Cell* **20**, 119-130. 2) Breuning *et al.* (1987) *Hum. Genet.* **76**, 287-289.

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A new *Hinc*II polymorphism at the epidermal growth factor receptor locus on chromosome 7

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Source/Description: A 2.4 kb cDNA (pE7) encoding part of the epidermal growth factor receptor was isolated from an A431 cDNA library by Xu *et al.* (1) and ligated into *Cla*I-digested pBR322.

Polymorphism: Digestion with *Hinc*II identifies a 2-allele polymorphism, with fragments of either 9.6 kb (A1) or 7.6 kb (A2) (See Figure 1). 9 smaller constant bands are also observed.

Frequency: Estimated from 19 unrelated Caucasians: A1: 0.71, A2: 0.29.

Estimated from 7 unrelated Peruvian Indians: A1: 0.71, A2: 0.29

Chromosomal Localisation: pE7 has been assigned to chromosome 7 p (2).

Mendelian Inheritance: Co-dominant segregation was demonstrated in 3 generations of one family, 13 individuals.

Probe Availability: American Type Culture Collection.

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References: 1) Xu, Y. *et al.* (1984) *Nature* **309**, 806-810. 2) Merlino, G.T. *et al.* (1985) *Molecular and Cellular Biology*, **5**, 1722-1734.

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