A *Dra*I RFLP detected for probe pIR4-3R [D15S11] on chromosome 15q

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Source and Description of Probe: pIR4-3R, a 0.4 kb fragment subcloned into pUC19 (1, 2), originally isolated from a flow-sorted library enriched for proximal 15q sequences.

Polymorphism: *Dra*I reveals a three allele polymorphism with band sizes 1.6 kb, 1.4 kb, 0.7 kb, and constant fragments at 1.8 kb and 0.3 kb.

Allele Frequency: Estimated from 27 unrelated Japanese.

C1: 1.6 kb allele: 0.72

C2: 1.4 kb allele: 0.18

C3: 0.7 kb allele: 0.10

Not Polymorphic For: *Bsp*1286I, *Cfr*13I, *Eco*0109I (studied in 6 unrelated individuals).

Chromosomal Localization: pIR4-3R has been assigned to 15q11.2-15q12, a segment missing in one #15 chromosome in 60% of Prader-Willi syndrome patients studied (1, 2, 3).

Mendelian Inheritance: Co-dominant segregation observed in 4 families with Prader-Willi syndrome patients.

Probe Availability: Available from ATCC.

References: 1) Donlon, T.A. *et al.* (1986) *Proc. Natl. Acad. Sci. USA* **83**, 4408-4412. 2) Tantravahi, U. *et al.* (1989) *Am. J. Med. Genet.* **33**, 78-87. 3) Hamabe, J. *et al.* (1991) *Am. J. Med. Genet.* in press.

An *Apa*I RFLP for the human porphobilinogen deaminase gene (PBGD)

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Source and Description of Probe: PUSE109, a 0.9 kb *Eco*RI-*Taq*I fragment subcloned into PGEM1, corresponding to the erythroid promoter and exon of the PBGD gene.

Polymorphism: *Apa*I reveals a two-allele polymorphism with band sizes 15.0 kb (D1) and 12.5 kb (D2).

Allele Frequency: Estimated from 40 unrelated Japanese.

D1: 15.0 kb allele: 0.11

D2: 12.5 kb allele: 0.89

Not Polymorphic For: *Ban*I, *Ban*II, *Bst*XI, *Rsa*I (studied in 8 unrelated individuals).

Chromosomal Localization: PBG-D has been assigned to 11q23-qter (2).

Mendelian Inheritance: Co-dominant segregation observed in 3 families with AIP.

Probe Availability: Information from M.Goosens (1).

References: 1) Llewellyn, D.H., Kalsheker, N.A., Elder, G.H. *et al.* (1987) *Nucl. Acids Res.* **15**, 1439. 2) Wang, A.L. *et al.* (1981) *Proc. Natl. Acad. Sci. USA* **78**, 5734-5738.