StyI polymorphism at the D15S11 locus

J.Hamabe and N.Niikawa
Department of Human Genetics, Nagasaki University School of Medicine, Sakamoto-Machi 12-4, Nagasaki 852, Japan

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: *Sty*I reveals a four allele polymorphism with band sizes 3.4 kb, 2.4 kb, 1.9 kb, 0.9 kb, and a constant fragment at 2.2 kb.

Allele Frequency: Estimated from 28 unrelated Japanese.
3.4 kb allele: 0.33
2.4 kb allele: 0.42
1.9 kb allele: 0.06
0.9 kb allele: 0.19

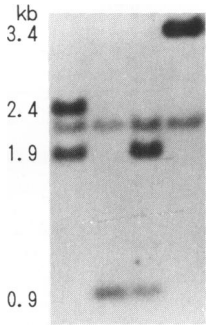
Not Polymorphic For: *Ban*II, *Hae*III, *Hin*cII, *Nci*I, *Nsi*I.

Chromosomal Localization: pIR4-3R has been assigned to 15q11.2-15q12, a segment missing in one #15 chromosome of a Prader-Willi syndrome patient with a prominent cytological deletion (1, 2).

Mendelian Inheritance: Codominant segregation observed in 8 families with Prader-Willi syndrome patients.

Probe Availability: Available from ATCC.

References: 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.



PstI RFLP of the CGB gene

M.Southall and G.Chenevix-Trench*
Queensland Institute of Medical Research and Joint Oncology Program, Department of Pathology, University of Queensland, Herston Medical School, Herston, Q 4006, Australia

Source and Description of Clone: pCGβ474 is a 580 bp cDNA clone of the beta sub-unit of the human chorionic gonadotrophin gene (1).

Polymorphism: PstI identifies two alleles:
A1-absence of 3.8 kb fragment
A2-presence of 3.8 kb fragment
Constant bands also at 3.0, 2.3, 1.7, 1.4, 1.2, 0.66 and 0.42 kb.

Frequencies: 14/28 unrelated Caucasians were of the 1,1 genotype.
Assuming Hardy-Weinberg equilibrium: A1 (-3.8 kb): 0.71
A2 (+3.8 kb): 0.29

Not Polymorphic For: EcoRI, HindIII, MspI, TaqI in 18 unrelated Caucasians.

Chromosomal Localization: CGβ and the beta chain of luteinizing hormone are part of a gene cluster at 19q13 (2).

Mendelian Inheritance: Mendelian inheritance was demonstrated in two two-generation families (Figure).

Probe Availability: Contact Dr Boime, Department of Pharmacology, Washington University Medical School, St. Louis, MO 63115.

Other Comments: 2,2 homozygotes cannot be distinguished from 1,2 heterozygotes even if the pCGβ474 plasmid is cut down to the 80 bp XmnI-HincII fragment which only detects the variable 3.8 kb fragment and the constant bands at 0.66 and 0.42 kb. This suggests that the RFLP may be due to an insertion/deletion.

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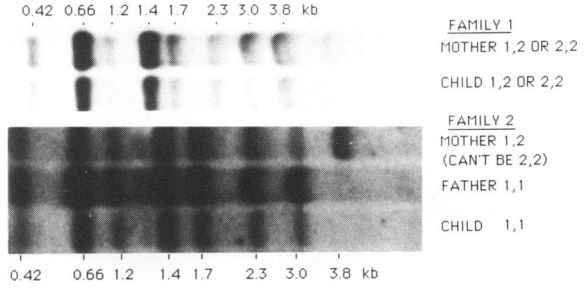
References: 1) Policastro *et al.* (1983) *J. Biol. Chem.* **258**, 11492-11499. 2) Brook *et al.* (1985) *Cytogenet. Cell Genet.* **40**, 590-591.



* To whom correspondence should be addressed