A third TaqI allele is detected by the probe pTD3-21 (D15S10) in Southern African chromosomes

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Source/Description: Probe pTD3-21 is a human cDNA clone which was isolated from HindIII total-digest libraries of flow-sorted inverted duplicated human chromosomes 15 (1) and has been subcloned as a 2.2 kb HindIII fragment in the vector pBR322. It maps to the region absent in Prader-Willi Syndrome patients with deletions (2).

Polymorphism: pTD3-21 was originally reported to detect a two-allele TaqI polymorphism with polymorphic fragment sizes of 9.0 kb and 8.2 kb and a constant band of 0.5 kb, with a PIC value of 0.28 (3). pTD3-21 has also been found to detect a third, African-specific allele of 3.0 kb.

Frequency: Determined in 20 random Caucasoid individuals, 23 random San individuals ('Bushmen') and 64 random Negroid individuals who are the parents of tyrosinase-positive oculocutaneous albino (ty-pos OCA) children.

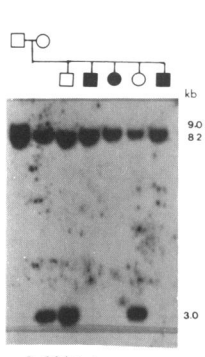
		Frequency		
Allele	Size (kb)	Negroid	San	Caucasoid
A1	9.0	0.48	0.15	0.90
A2	8.2	0.40	0.52	0.10
A3	3.0	0.12	0.33	–
	PIC	0.59	0.60	0.16

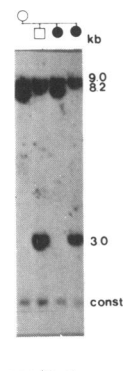
Chromosomal Localisation: Probe pTD3-21 (D15S10) maps to the region 15q11-q12 (1).

Mendelian Inheritance: Codominant segregation has been shown in 9 families, two of which are shown below. The black symbols represent children with ty-pos OCA which is inherited as an autosomal recessive.

Probe Availability: Probe pTD3-21 was purchased from ATCC (depositor Marc Lalande).

References: 1) Donlon *et al.* (1986) *PNAS USA* **83**, 4408-4412. 2) Latt *et al.* (1987) *Cytogenet. Cell Genet.* **46**, 644. 3) Nicholls *et al.* (1989) *Am. J. Med. Genet.* **33**, 66-77.





Microsatellite polymorphism at the D9S12 locus

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Source/Description: Cosmid T512 — which we obtained by screening a human genomic library with a HinfI fragment of the probe pTHH22 (Holm *et al.*, 1988) — contains $(CA)_{16}$, the flanks of which were sequenced as described (Yuille *et al.*, 1991). Alleles were typed using the polymerase chain reaction.

Primer Sequences:
5′ CCT CCA CAT GGA CTC ACC TG 3′ (CA strand);
5′ AAG GGG AGG GAA TCA GGT GT 3′ (TG strand).

Polymorphism: Heterozygosity was estimated at 74% by analysis of 92 chromosomes from unrelated individuals.

Allele	bp	Frequency
B1	194	0.01
B2	192	0.05
B3	190	0.10
B4	188	0.05
B5	186	0.06
B6	184	0.21
B7	182	0.43
B8	180	ND
B9	178	0.01
B10	176	0.08

Chromosomal Localisation: Fluorescence *in situ* hybridisation localises cosT512 to Chr 9q22.31.

Mendelian Inheritance: Co-dominant segregation was observed in 9 families of 2, 3 and 4 generations.

Comments: Alleles were typed as described (Yuille *et al.*, 1990) except: all NTPs were at 125 μM; 1.0 μCi of alpha ^{32}P-dCTP was used; samples were heat denatured at 95°C and snap-chilled on ice before gel-loading. EMBL Data Library accession number for sequence data is X60736.

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References: 1) Yuille,M.A.R. *et al.* (1990) *Nucl. Acids Res.* **18**, 7472. 2) Yuille,M.A.R. *et al.* (1991) *Nucl. Acids Res.* **19**, 1950. 3) Holm,T. *et al.* (1988) *Nucl. Acids Res.* **15**, 5216.