A new polymorphic probe on chromosome 22: NB5 (D22S201)

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Source/Description: NB5 is a 0.5 kb HindIII-EcoRI fragment isolated from two pooled chromosome 22 specific libraries (ATCC # 57733 and ATCC # 57714) and was subcloned into pUC9.

Polymorphism: TaqI digestion of genomic DNA and hybridization with the probe detects a four allele polymorphism: 1.75 kb (A1), 1.6 kb (A2), 1.4 kb (A3) and 1.3 kb (A4). No constant bands were present.

Frequency: Estimated from 97 unrelated Caucasians.
A1 : 0.072
A2 : 0.86
A3 : 0.052
A4 : 0.015

Not Polymorphic For: BglI, BglII, DraI, MspI and PstI.

Chromosomal Localization: Regional localization was established by hybridization to a panel of somatic cell hybrids: PgMe-25Nu, containing only human chromosome 22; PgMo-22 and 1CB-17ANu, respectively containing both products of the Philadelphia translocation at 22q11; A3EW2-3B, containing the t(11;22) from Ewing's sarcoma (at 22q12) and 1/22 AM27 containing a t(1;22) at 22q13 (1, 2). The probe was assigned to chromosome 22 between 22q13 and 22qter.

Mendelian Inheritance: Mendelian inheritance has been demonstrated in extended pedigrees of Gilles de la Tourette syndrome families (n = 380).

Probe Availability: Available for collaboration.

References: 1)Goyns,M.H., Young,B.D., Geurts van Kessel,A., de Klein,A., Grosveld,G., Bartram,C.R. and Bootsma,D. (1984) *Leukemia Res.* **8**, 547–553. 2)Geurts van Kessel,A., Turc-Carel,C., de Klein,A., Grosveld,G., Lenoir,G. and Bootsma,D. (1985) *Mol. Cell Biol.* **5**, 427–429.

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Two microsatellite polymorphisms at the D5S39 locus

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Source/Description: A human genomic library in EMBL3 was screened with a probe of p105–153Ra (D5S39) (1). A positive clone, lambda153–6741, was isolated. Two AluI fragments hybridised to poly (dC-dA). poly (dG-dT) and designated 153–6741GT and 153–6741CA. Primers were designed to amplify the repeats.

PCR Primers:
153–6741GT 1 5'-CCATTGTATTAGGGTTCTCCAG-3'
153–6741GT 2 5'-CTCTTGGTTTCCTGGCTTCGG-3'
153–6741CA 1 5'-GCTCCAAGGTAAATGCCAGAC-3'
153–6741CA 2 5'-CTGGAGAACCCTAATACAATGG-3'

Frequency: Estimated from 54 (153–6741GT) and 30 (153–6741CA) chromosomes of unrelated European Caucasians.

	153-6741GT			153-6741CA	
Allele	Size (nt)	Frequency	Allele	Size (nt)	Frequency
1	220	0.29	A	124	0.57
2	218	0.25	B	122	0.43
3	216	0.22			
4	214	0.12			
5	212	0.12			
	Heterozygosity = 78%			Heterozygosity = 47%	

Chromosomal Localisation: Pairwise linkage analysis between 153–6741GT and D5S39 (1) in 7 families gave a maximum lod of 17.175, theta = 0, indicating localisation to chromosome 5q12-q14. 153–6741CA and D5S39 gave a maximum lod of 14.809, at theta = 0.

Mendelian Inheritance: Co-dominant segregation was observed in 12 two or three generation families.

PCR Conditions: 25 μl containing: 50 ng DNA, 5 pmoles each primer, 1.5 mM $MgCl_2$, 10 mM Tris-Cl pH 8.3, 50 mM KCl, 25 μM dATP and 200 μM other dNTPs, 1 unit Taq polymerase, 0.01% gelatin, 1 μl of ^{35}S-dATP at 500 Ci/mmol (Dupont). 30 cycles with denaturation at 94°C, annealing at 60°C (153–6741GT) or 59°C (153–6741CA) extension at 72°C, all for 1 minute each.

Other Comments: Using ASSOC (2). 153–6741CA and 153–6741GT showed significant allelic association with the MspI RFLP at D5S39 and with each other. However, p-values are unreliable due to the small expected cell probabilities. Highly significant association was found between 153–6741CA and the MspI RFLP with a Fisher's exact test.

Acknowledgements: Wellcome Trust, Medical Research Council, Merck, Sharpe and Dohme.

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