# A highly polymorphic cDNA probe in the *NF1* gene

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*Source and Description*: A 2.3 kb cDNA clone, AE25 (1), obtained by screening a human fetal brain cDNA library (Stratagene, catalog number 936206), with P5, a cDNA clone in the 3′ translated region of the neurofibromatosis type 1 (NF1) gene (2). The insert was subcloned into the EcoRI site of Bluescript.

*Polymorphisms and Frequencies*: Digestion of genomic DNA with one of four enzymes reveals polymorphism, listed schematically below. There appears to be strong linkage disequilibrium between the different RFLP's, particularly between the polymorphisms identified by BglII and PvuII.

*Not Polymorphic For*: HincII and XbaI (studied in 7 and 19 individuals, respectively).

*Chromosomal Localization*: This probe is part of the NF1 gene on 17q11.2, and represents nucleotides 3685–6012 of the published cDNA sequence (3).

*Mendelian Inheritance*: Co-dominant segregation of the PvuII, TaqI, and BglII polymorphisms demonstrated in 3 three generation families (15 meioses).

*Clinical Relevance*: Linkage analysis in families with neurofibromatosis type 1 (NF1).

*Probe Availability*: Apply to Francis S.Collins.

*Acknowledgements*: This work was supported by NIH grant NS 23410 to FSC and a fellowship from University of Copenhagen, Denmark, to LBA. FSC is an investigator of the Howard Hughes Medical Institute.

*References*: 1) Marchuk,D. *et al.*, in preparation. 2) Wallace,M. *et al.* (1990) *Science* **249**, 181–186. 3) Xu,G. *et al.* (1990) *Cell* **62**, 599–608.

**Table**

| enzyme | # of individuals studied | allele frequencies | allele sizes |
|---|---|---|---|
| PvuII | 39 | A: 0.41 | A: 7.2 kb |
| | | B: 0.51 | B: 6.8 kb |
| TaqI | 28 | A: 0.57 | A: 7.1 kb |
| | | B: 0.43 | B: 6.7 kb |
| BglII | 25 | A: 0.50 | A: 4.2 kb |
| | | B: 0.50 | B: 3.8 kb |
| MspI | 16 | A: 0.03 | A: 2.8 kb |
| | | B: 0.97 | B: 2.6 kb |

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# CA repeat polymorphism in the glucose transporter GLUT 2 gene

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*Source/Description*: The polymorphic (CA)15 is located in intron 4a of human glucose transporter gene 2 (GLUT2). Two oligonucleotides flanking the repeat sequence were used to selectively amplify the sequence from genomic DNA employing polymerase chain reaction (PCR). The predicted length of the amplified sequence is 104 bp.

*Primer Sequence*:
TG primer 5′-GCTGGAAGAAGCATATCAGG-3′
CA primer 5′-CTTTACTTGAGATTTCCGCC-3′

*Polymorphism*: Six alleles were detected.

*Frequency*: Estimated from 156 chromosomes of unrelated European Caucasian individuals. Heterozygosity: 76%.

| Allele | Length (bp) | Number of (CA) Repeats | Frequency |
|---|---|---|---|
| K1 | 114 | 20 | 0.04 |
| K2 | 112 | 19 | 0.14 |
| K3 | 110 | 18 | 0.19 |
| K4 | 108 | 17 | 0.47 |
| K5 | 106 | 16 | 0.04 |
| K6 | 104 | 15 | 0.14 |

*Mendelian Inheritance*: Autosomal co-dominant segregation was observed in 20 families with 167 individuals.

*Chromosomal Localization*: Glucose Transporter Gene 2 (GLUT2) has been assigned to chromosome 3q26.1–3q26.3 (1).

*Other Comments*: The PCR reaction was as previously described (2) using end-labeled oligonucleotide OL1 (TG primer) and unlabeled OL2 (CA primer). Each 50 μl reaction contained 400 ng genomic DNA, 140 ng (10.6 picomoles) each primer, 1.25 U Taq DNA Polymerase, 200 uM dNTP. Amplification conditions were 94°C/7 mins, then 25 cycles of 94°C/1 min, 60°C/1 min, and a final elongation step of 73°C/7 mins. A 3 μl aliquot was electrophoresed on a 6% polyacrylamide sequencing gel which was fixed, dried and exposed to Amersham RX Film.

*References*: 1) Fukumoto,H. *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 5434–5438. 2) Saiki,R.K. *et al.* (1988) Science **239**, 487–491.

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