A new polymorphic probe on chromosome 3p: λ LIB36-68 (D3S615)

L.Geil¹, F.Latif, M.Yao, M.L.Orcutt, B.Zbar and M.I.Lerman*

Laboratory of Immunobiology, National Cancer Institute, and ¹Biological Carcinogenesis and Development Program, Program Resources Inc., DynCorp, Frederick Cancer Research and Development Center, Frederick, MD 21701, USA

Source/Description: λ LIB36-68 is a 3.8 kb EcoRI fragment isolated from a Charon 21A human chromosome 3 library (1).

Polymorphism: DraI digestion of genomic DNA and hybridization with the probe detects a two-allele polymorphism: 1.8 kb (A1) and 1.5 kb (A2).

Frequency: Estimated from 120 unrelated Caucasians
A1:0.82
A2:0.18

Frequency of Heterozygosity: 0.30.

Not Polymorphic For: BamHI, BglII, DraI, EcoRI, HindIII, HinfI, MspI, PvuII and TaqI.

Chromosomal Localization: Using a somatic cell hybrid panel (2) which was based on linkage groups anchored to physically localized markers, the probe was assigned to the short arm of chromosome 3 between 3p21-p25.

Mendelian Inheritance: Mendelian inheritance has been demonstrated in a two generation von Hippel-Lindau disease (VHL) family.

Probe Availability: Available for collaboration.

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References: 1) American Type Culture Collection No. 57717, National Lab ID Code LA03NS01. 2) Lerman, M., Latif, F., Glenn, G. *et al.* (1991) Isolation and regional localization of a large collection (2,000) of single copy DNA fragments on human chromosome 3 for mapping and cloning tumor genes. *Human Genetics* **86**, 567-577.

Dinucleotide repeat polymorphism in Gs- α subunit gene (*GNAS1*) on chromosome 20

M.Granqvist, K.Xiang, M.Seino and G.I.Bell*

Howard Hughes Medical Institute, University of Chicago, 5841 S. Maryland Ave, Box 391, Chicago, IL 60637, USA

Primers/Description: Two primers (GNAS1-A, 5'-ACGTTAGC-TTGGTGGATAAG-3', and GNAS1-B, 5'-AGAGCTAGAGTGGTTACCTG-3') were used to amplify a 217-220 bp CA repeat-rich region located in intron 3 of the human Gs- α subunit gene (*GNAS1*) (1).

Frequency: Three alleles were observed in 43 unrelated Caucasians. The heterozygosity was 58%.

Allele (bp)	Frequency
B1 220	0.59
B2 218	0.40
B3 217	0.01

Chromosomal Localization: *GNAS1* was assigned to the long arm of chromosome 20 (2).

Mendelian Inheritance: Co-dominant inheritance was observed in a five-generation pedigree in which 75 individuals were typed.

Other Comments: The PCR was performed using ³²P-labeled GNAS1-A and unlabeled GNAS1-B for 30 cycles: denaturation at 94°C for 1 min; annealing at 55°C for 1 min; and extension at 72°C for 2 min. The PCR products were analyzed on a 5% denaturing polyacrylamide gel (Fig. 1). The sequence of the C-A-repeat rich region of *GNAS1* has been submitted to GenBank and is also available from the authors.

References: 1) Kozasa, T. *et al.* (1988) *Proc. Natl. Acad. Sci. USA* **85**, 2081-2085. 2) Gejman, P.V. *et al.* (1991) *Genomics* **9**, 782-783.

Figure 1. PCR amplification of *GNAS1* dinucleotide repeat DNA polymorphism. The alleles are noted at the right and the genotypes at the bottom of the figure.

* To whom correspondence should be addressed

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