Identification of an insulin receptor exon 8 *NsiI* polymorphism using the polymerase chain reaction

Craig L.Hanis* and Terry K.Bertin

Center for Demographic and Population Genetics,
Graduate School of Biomedical Sciences, University of
Texas Health Science Center Houston, PO Box 20334,
Houston, TX 77225, USA

Source/Description: Regions of the human insulin receptor (INSR) locus have been sequenced on multiple occasions and a potential polymorphism in exon 8 postulated based on sequence differences (a synonymous substitution of A for G in the third position of amino acid codon 523; nucleotide 6244) (1). Examination of the sequence differences indicated that restriction enzyme *NsiI* would distinguish the two postulated forms. Exon 8 was amplified from NaCl extracted DNA using oligonucleotides and conditions recently reported (2).

Polymorphism: Digestion with *NsiI* confirmed that this site is, in fact, polymorphic. Allele N1 (sequence ATGGGT at 6240–6245) is not cut and yields a 322 bp fragment. Allele N2 (ATGCAT) has the cut site and gives fragments of length 238 bp and 84 bp. All fragments can be visualized directly on ethidium stained 1% agarose gels.

Frequency: Frequencies in a random sample of 179 Mexican-Americans from Starr County, Texas were: $f(N1) = .765$, $f(N2) = .235$ with no evidence for departures from Hardy-Weinberg expectations.

Not Polymorphic For: Not applicable.

Chromosomal Localization: The insulin receptor maps to chromosome 19p13.3–13.2.

Mendelian Inheritance: Codominant Mendelian segregation was demonstrated in 24 offspring in 7 nuclear families.

Probe Availability: Not applicable.

Other Comments: Adequate restriction was obtained following a minimum 6-hour incubation at 37°C without the addition of restriction buffer.

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References: 1) Moller, D.E., Yokota, A. and Flier, J.S. (1989) *Diabetes* **38**, 1496–1500. 2) Seino, S., Seino, M. and Bell, G.I. (1990) *Diabetes* **39**, 123–128.

TaqI polymorphism at the D6S91 locus

Huda Y.Zoghbi¹ and Alanna E.McCall

Department of Pediatrics and ¹Institute for Molecular
Genetics, Baylor College of Medicine, Houston, TX
77030, USA

Source/Description: pHZ-49 is a 2.7 kb fragment with *EcoRI* ends cloned in pBluescript II KS. It was subcloned from λHZ-49 which was isolated from a genomic library constructed in λDASH from a somatic cell hybrid retaining chromosome 6p as the only human chromosome (1).

Polymorphism: *TaqI* identifies a two allele polymorphism 5.1 kb (A1) and 4.8 kb (A2). No constant bands were present.

Frequency:

35 unrelated Caucasians: A1 = 0.79 A2 = 0.21

7 unrelated U.S. Blacks: A1 = 0.35 A2 = 0.65

Not Polymorphic For: *ApaI*, *BamHI*, *BanI*, *BanII*, *BclI*, *BglI*, *BglII*, *DraI*, *EcoRI*, *EcoRV*, *Fnu4HI*, *HaeIII*, *HindIII*, *HinfII*, *MobI*, *PstI*, *PvuII*, *RsaI*, *ScaI*, *SstI*, *XbaI*.

Chromosomal Localization: pHZ-49 localizes to 6p11–6p21 using a previously published chromosome 6 somatic cell hybrid panel (1, 2).

Mendelian Inheritance: Mendelian inheritance was observed in 17 CEPH families and in 54 individuals from a seven generation spinocerebellar ataxia family.

Probe Availability: Contact H.Y.Zoghbi.

Other Comments: Prehybridization with human placental DNA is required prior to hybridization at normal stringency (65°C, 1M Na⁺).

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References: 1) Zoghbi et al. (1990) *Genomics* **6**, 352. 2) Zoghbi et al. (1988) *Genomics* **3**, 396.

* To whom correspondence should be addressed