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# Dinucleotide repeat polymorphism at the RYR1 locus (19q13.1)

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*Source/Description*: A HaeIII fragment from a cosmid containing a portion of the human skeletal Ryanodine receptor gene (RYR1) (1) was selected by hybridisation to poly(dC-dA).poly(dG-dT) and was designated Wis1. Sequencing was performed for determination of primer sequences for the polymerase chain reaction. The predicted length of the amplified fragment was 227 bp.

*Primer Sequences*:
5'GCATCACGGTCTGCAATTCAT 3' (CA strand);
5'GCAATGGCATAATCTCAGCT 3' (GT strand).

*Frequency*: Estimated from 160 chromosomes of the CEPH parents.
Observed heterozygosity = 0.43, PIC = 0.38

| Allele (bp) | Frequency | Allele (bp) | Frequency |
|---|---|---|---|
| A1 233 | 0.0125 | A5 225 | 0.0250 |
| A2 231 | 0.0125 | A6 223 | 0.0313 |
| A3 229 | 0.1000 | A7 221 | 0.0500 |
| A4 227 | 0.7688 | | |

*Chromosomal Localisation*: The skeletal muscle Ryanodine receptor gene (RYR1) has been mapped to 19q13.1 (2).

*Mendelian Inheritance*: Co-dominant segregation was observed in 40 two generation families.

*PCR Conditions*: Amplification reactions were as follows, denaturation at 94°C (3 min), 32 cycles of denaturation at 94°C (1 min), annealing at 54°C (45 sec) and extension at 72°C (1 min) using 0.5 U Amplitaq, 1×GeneAmp buffer and 200 μM dNTPs (Perkin Elmer-Cetus), 100 ng genomic DNA and 10 pmol of each primer in a volume of 15 μl. Allele sizes were determined on 8% denaturing polyacrylamide gels with autoradiography by comparison to the cloned sequence. The sequenced repeat was (CA)17. The sequence has been submitted to Genbank. Accession number = X60187 Human RYR1 DNA.

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*References*: 1) Zorzato,F. *et al.* (1990) *J. Biol. Chem.* **265**, 2244–2256. 2) MacKenzie,A.E. *et al.* (1990) *Am. J. Hum. Genet.* **46**, 1082–1089.

# Microsatellite polymorphism in human insulin receptor gene (*INSR*) on chromosome 19

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*Primers/Description*: Two primers (INSR E3–2B, 5'-ATTGC-TGCATATGCAGACAG-3' and INSR E3-rC3, 5'-TGC-AGCCGTGTGACTTACAG-3') were used to amplify a 138–156 bp ATTT and CT repeat-rich region in intron 2 of the human *INSR* gene (1).

*Frequency*: Nine alleles were observed in 48 unrelated Caucasians. The heterozygosity was 58%.

| Allele | bp | Frequency | Allele | bp | Frequency |
|---|---|---|---|---|---|
| AA1 | 156 | 0.01 | AA2 | 154 | 0.01 |
| AA3 | 152 | 0.01 | AA4 | 150 | 0.10 |
| AA5 | 148 | 0.65 | AA6 | 146 | 0.05 |
| AA7 | 144 | 0.03 | AA8 | 142 | 0.12 |
| AA9 | 138 | 0.02 | | | |

*Chromosomal Localization*: *INSR* was assigned to chromosome 19p13.3-p13.2 (2).

*Mendelian Inheritance*: Codominant inheritance was observed in four nuclear families.

*Other Comments*: The PCR was performed using $^{32}$P-labeled INSR E3–2B and unlabeled INSR E3-rC3 for 30 cycles: denaturation at 94°C for 1 min; annealing at 56°C for 2 min; and extension at 72°C for 2.5 min. The PCR products were analyzed on a 5% denaturing polyacrylamide gel (Fig. 1). The microsatellite sequence in intron 2 was of the form $(ATTT)_{11}CC(CT)_{10}$.

*References*: 1) Seino,S. *et al.* (1990) *Diabetes* **39**, 123. 2) Yang-Feng,T.L. *et al.* (1985) *Science* **228**, 728.

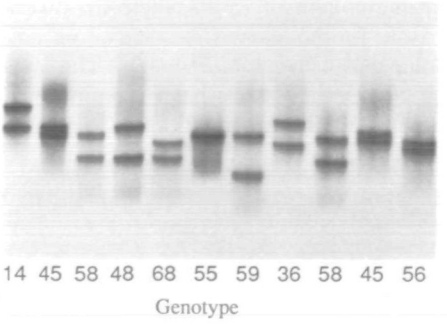



**Figure 1.** PCR amplification of microsatellite repeat polymorphism in *INSR* gene. The genotypes are noted at the bottom of the figure.

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