Trinucleotide repeat polymorphism at the human insulin-like growth factor I receptor gene (IGF1R)

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Source/Description: The polymorphism is a trinucleotide repeat of the form (AGG)₇ and starts at position 967 of the human insulin-like growth factor I receptor gene on chromosome 15q25-qter (GenBank accession no. M69229) (1). The polymorphism can be typed using the polymerase chain reaction (PCR) as previously described (2). The predicted size of the amplified sequence is 96 bp.

Primer Sequences:

GCTGAGGGAGGAGGCGGC (CA strand)
GGCGAGGGGCAGAAACGC (TG strand)

Frequency: Estimated from 90 chromosomes of unrelated CEPH individuals. Observed heterozygosity is 44% with a PIC of 0.35.

Allele	(bp)	Frequency
C1	96	0.01
C2	93	0.69
C3	90	0.30

Mendelian Inheritance: Co-dominant segregation was observed in three large CEPH families.

Chromosomal Localization: The human IGF1R gene has been assigned to chromosome 15q25-qter (3).

PCR Conditions: The PCR reaction was performed using 200 ng genomic DNA, 10 pMoles of each primer, 1.5 units of Taq polymerase and 2.5% formamide in a final volume of 50 μ l. Samples were subjected to 30 cycles consisting of 40 seconds at 92°C, 30 seconds at 63°C and 30 seconds at 72°C.

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Trinucleotide repeat polymorphism in the androgen receptor gene (AR)

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Source/Description: The human androgen receptor gene contains in exon one a (CAG)_N(CAA) repeat, encoding a polyglutamine stretch, at position 172 following the translation start codon (1). This polymorphism is extremely useful for diagnosis in families with the Androgen Insensitivity Syndromes, which display a high incidence of distinctive mutations in the androgen receptor gene (2–5). The polymorphism could be typed using the polymerase chain reaction (PCR). The length of the repeat in allele B10 was (CAG)₁₉(CAA).

Primer Sequences:

Forward 5'TCCGCGAAGTGATCCAGAAC
Reverse 5'CTTGGGGAGAACCATCCTCA

Frequency: Eleven different allele products were detected among 63 chromosomes of 22 female and 19 male, unrelated Caucasians. The heterozygosity in females is 0.89.

Allele	(bp)	Frequency	Allele	(bp)	Frequency
B11	195	0.03	B5	213	0.13
B10	198	0.17	B4	216	0.11
B9	201	0.11	B3	219	0.03
B8	204	0.16	B2	222	0.03
B7	207	0.08	B1	225	0.02
B6	210	0.13			

Chromosomal Location: The androgen receptor gene (AR) has been assigned to Xq11–12 (6).

Other Comments: 200 ng of genomic DNA was amplified using 60 ng of each primer and 0.02 U Super Taq DNA polymerase (HB Technology, Cambridge) in a total volume of 15 μ l in a buffer, containing 50 mM KCl, 1.5 mM MgCl₂, 0.1% Triton X-100, 0.2 mM of dATP, dCTP, dGTP and TTP each, and 10 mM Tris-HCl (pH 8.8), supplemented with 2.5 μ Ci [α -³²P]dATP. Conditions for amplification: 30 cycles of 1 minute 95°C, 2 minutes 55°C, and 2 minutes 72°C. The final elongation cycle was 10 minutes at 72°C. The radioactive amplification products were analyzed on a 6% denaturing polyacrylamide sequencing gel.

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