SphI RFLP at the human growth hormone gene cluster

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Source/Description: The probe used was an EcoRI-HindIII fragment (position 36414−36587) of the human growth hormone (hGH) locus (filled rectangle in Fig. 1). Duplications of this sequence are also present at positions 21632−21804 and 59843−60016 of the locus (unfilled rectangles in Fig.1) (1).

Polymorphism: SphI detects a two allele polymorphism with bands of >23 (J1) and 4.1 kb (J2). In addition, two constant bands of 6.0 and >23 kb are observed.

Frequency: Estimated in 24 unrelated individuals.
J1: 0.65
J2: 0.35

Chromosomal Localization: 17q22−24 (1).

Mendelian Inheritance: Confirmed in two informative families.

Probe Availability: Contact G.J.

Other Comments: RFLPs for BamHI, BglII, HincII and MspI have been previously identified at the hGH locus (2). The actual size of the J1 allele is 31.6 kb (1). The >23 kb constant band is a heterogeneous fragment which is 27.5 kb in size in the J2 haplotype and corresponds to the J1 allele in the J1 haplotype. In the heterozygote, the >23kb band is a mixture of both the 27.5 and the 31.6 kb fragments.

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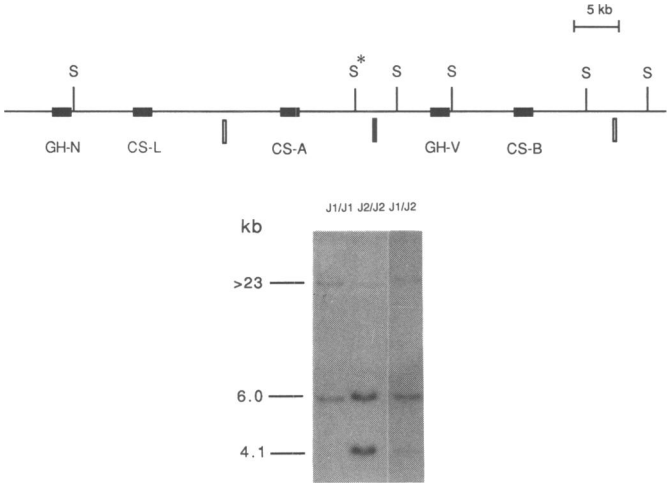

Dinucleotide repeat polymorphism at the PGK1P1 locus

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Source and Description of Clone: The $(CA)_{14}$ repeat region in cosmid ICRFc100F0599 (EMBL accession number X56251) has been previously described (1).

PCR Primers:
PGK/5A = 5′-TCTACATGTACTTAACCTGC-3′
PGK/5B = 5′-AACTCAGTTTTGAGCTCCTA-3′

Polymorphism, Frequencies, Mendelian Inheritance and PCR Conditions: See reference (1).

Chromosomal Localization: We originally placed this repeat at the PGK1 locus at Xq13 (1) because the cosmid ICRFc100F0599 hybridized with the PGK1 probe pSPT-PGK and no recombinants between the $(CA)_n$ repeat and the PGK PstI RFLP were seen in 35 phase known meioses in 4 CEPH families. We have now obtained physical and genetic evidence which places ICRFc100F0599 at the closely linked pseudogene locus PGK1P1 at Xq12 (2). 1.) The published pseudogene sequence (3) was used to design PCR primers which specifically amplify this pseudogene sequence. These primers amplified a single fragment from genomic DNA and ICRFc100F0599; the amplified fragment had the predicted size and contained restriction sites predicted from the unpublished sequence. 2.) Two individuals from different families affected with X-linked hypohydrotic ectodermal dysplasia had recombinant PGK1P1 $(CA)_n$/PGK PstI RFLP haplotypes.

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