RFLPs of the gene for the human glycine receptor on the X-chromosome

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SOURCE/DESCRIPTION: A 650bp EcoRI fragment from the 5' end of the cDNA clone of the gene for 48kd polypeptide subunit of the human glycine receptor (unpublished data). This clone was obtained by screening a human cDNA library with a rat glycine receptor probe (1). Vector: pUC18.

POLYMORPHISM: HincII recognizes two alleles with bands at 5.5 kb (A₁) and 3.0 kb (A₂). BstNI and BanI recognize two alleles with bands at 1.4 kb (B₁) and 1.35 kb (B₂) and 5.5 (C₁) and 3.0 kb (C₂) respectively. Six invariant bands are seen with each enzyme.

FREQUENCY: HincII A ₁ (5.5 kb):0.44	BstNI B ₁ (1.45 kb):0.34
A ₂ (3.0 kb):0.56	B ₂ (1.35 kb):0.66
Studied in 29 unrelated caucasians.	BanI C ₁ (4.8 kb):0.66
	C ₂ (4.7 kb):0.34 Studied in 6 individuals.

NOT POLYMORPHIC FOR: BamHI, BglII, BglIII, EcoRI, EcoRV, HincIII, HinfI, MspI, PstI, PvuII, RsaI, SacI, StuI, TaqI, XbaI, and XmnI.

CHROMOSOMAL LOCALISATION: The gene for the 48Kd polypeptide of the human glycine receptor has been mapped to the Xp21.2-Xp22.1 region of the X-chromosome by hybrid cell analysis (unpublished data).

MENDELIAN INHERITANCE: X-linked transmission was demonstrated in 3 separate families with no male to male transmission in 8 informative matings.

PROBE AVAILABILITY: The human glycine receptor probe is available for linkage analysis from Dr. Heinrich Betz at the above address.

OTHER COMMENTS: This probe is from the 5' end of the gene for the 48Kd polypeptide of the human glycine receptor. The genes for the 58KD & 93KD proteins have not been cloned yet. Additional bands localize to autosomes, suggesting more than one loci for the glycine receptor.

REFERENCE: 1. Grenningloh G., Rienitz A., Schmitt B., Methfessel C., Zensen M., Beyreuther K., Gundelfinger ED., Betz H.: The Strychnine-Binding Subunit of the Glycine Receptor Shows Homology with Nicotinic Acetylcholine Receptors. Nature (1987) 328:215-220.