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RFLPs in human X-linked PGK1: a new probe for the PstI RFLP demonstrates strong linkage disequilibrium with the BglI RFLP

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INTRODUCTION: Several RFLP's have been described in the human phosphoglycerate kinase 1 (PGK1) gene in Xq13. Whereas the PstI polymorphism as detected with PGK1 cDNA (Hutz et al.) was complicated by multiple X-linked and autosomal constant bands, only two bands are seen with a fourth-intron genomic probe. We have compared this PstI RFLP to the 5' BglI RFLP detected by pSPT-PGK (Vogelstein et al.) and have found them to be in strong linkage disequilibrium.

SOURCE AND DESCRIPTION OF CLONE: pXPGK-RI0.9, provided by Dr. Stuart Orkin, is a genomic fragment cloned into the EcoRI site of pBR322; it contains the last 83 bp of exon 4 and extends 820 bp into the fourth intron.

POLYMORPHISM AND FREQUENCIES: pXPGK-RI0.9 detects PstI fragments of 5.2kb (A1) or 1.8kb (A2), and, occasionally, a faint 5.2kb autosomal band (figure, lane 5). We found 85% of 99 unrelated X chromosomes with A1 and 15% with A2 (in contrast to the 60% A1 originally reported by Hutz). These allele frequencies are close to the BglI RFLP frequencies of 21% B1 and 79% B2 reported by Vogelstein and observed in our study of 170 unrelated X chromosomes.

AVAILABILITY: Will be available through ATCC.

COMPARISON OF 4TH INTRON PST I AND 5' BGL I RFLP'S: 86 male and phase-known X chromosomes tested for both RFLPs showed 87% of maximal possible disequilibrium, with only one A1,B1 (figure, lane 8) and one A2,B2 (figure, lane 7).

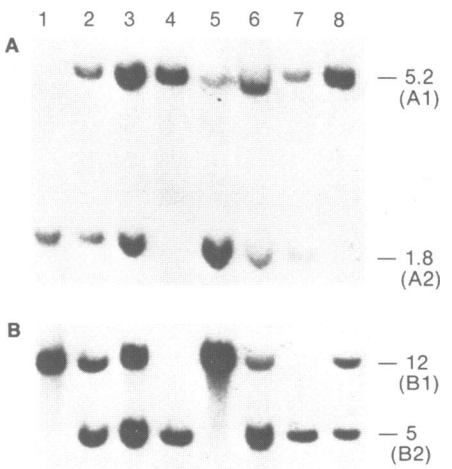

FIGURE: Southern blots illustrating the alleles of eight females as detected by (A) pXPGK-RI0.9 and (B) pSPT-PGK. Lanes 1 and 5: A2,B1 homozygotes; lane 5, which is overloaded, shows the faint 5.2 kb autosomal band. Lane 4: A1,B2 homozygote. Lanes 2, 3 and 6: double heterozygotes. Lane 7: A1,B2 and A2,B2 (rare). Lane 8: A1,B2 and A1,B1 (rare).

REFERENCES:
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Vogelstein. 1987. Ca Res 44:4806-4813.

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