Nsi I RFLP at the X-linked chronic granulomatous disease locus (CYBB)

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SOURCE AND DESCRIPTION OF CLONE: This cDNA probe is a PstI-KpnI fragment of approximately 3.5 kb which encompasses the majority of the transcript of the β-subunit of cytochrome b_{245} in a gemini 4 plasmid (1).

POLYMORPHISMS: Nsi I reveals invariant fragments of: 7.7, 6.4, 4.1, 2.8 and 0.95 kb and polymorphic fragments of 1.7 kb (allele A1) and 1.3 kb (allele A2) (Figure 1).

FREQUENCIES: Sixteen unrelated Causasians, 13 females and 3 males, were tested. The 1.7 kb A1 allele was present in 21/29 (0.72) and the 1.3 kb A2 allele in 9/29 (0.18) X chromosomes.

NOT POLYMORPHIC FOR: BamHI, BanII, Dra I, Eco 0109, EcoRI, HindIII, HindIII/MluI, PstI, PvuII, RsaI, SacI, TaqI, XmnI in 10-15 X chromosomes tested.

CHROMOSOMAL LOCALIZATION: The CGD locus has been assigned to Xp21.1 (2-3)

MENDELIAN INHERITANCE: X-linked inheritance of the NsiI RFLP was observed in two three generation families.

PROBE AVAILABILITY: Available from S.H. Orkin, Boston Childrens Hospital.

REFERENCES: (1) Royer-Pokora et al. (1986) Nature 322, 32-38.
(2) Francke (1984) Cytogenet. Cell Genet. 38, 298-307.
(3) Francke et al. (1985) Amer. J. Hum. Genet. 37, 250-267.

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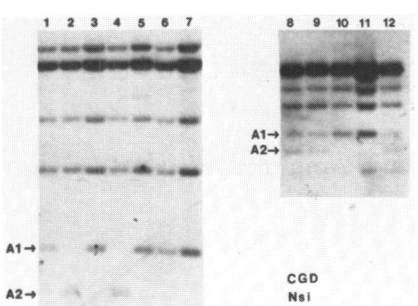

Figure 1
Lanes 1, 2, 4 and 6: Males
Lanes 3, 5, 7 and 12: Females