An additional NsiI RFLP at the X-linked chronic granulomatous disease (CYBB) locus

A. Pelham, S. Malcolm, R. J. Levinsky and C. Kinnon

Departments of Immunology and Genetics, Institute of Child Health, 30 Guilford Street, London WC1N 1EH, UK

Description of Probe: A cDNA probe of about 3.5 kb which is a PstI-KpnI fragment encoding the majority of the beta chain of cytochrome b-245 transcript in pGEM-4 (1).

Polymorphisms: Digestion with NsiI reveals invariant fragments of approximately 7.2, 6.2 and 3.7 kb. In addition to the polymorphic fragments of 1.7 and 1.3 kb described by Battat and Francke (2) a further polymorphism exists with fragment sizes of 2.9 and 2.5 kb.

Frequency: In 32 unrelated chromosomes, allele frequencies were 2.9 kb: 19% and 2.5 kb: 81%. In 29 of the same chromosomes allele frequencies were 1.7 kb: 90% and 1.3 kb: 10%. The observed haplotype distribution does not deviate significantly from that expected on the basis of the above allele frequencies, implying that there is no strong linkage disequilibrium.

Not Polymorphic For: BamHI, BglII, EcoRI, HindIII, MspI, PstI, TaqI in 14 chromosomes tested.

Chromosomal Localisation: The X-linked CYBB locus has been assigned to Xp21.1 (1).

Mendelian Inheritance: X-linked inheritance has been demonstrated in 2 three-generation families.

Probe Availability: Available from S.H.Orkin, Boston Children's Hospital.

Acknowledgements: We thank S.H.Orkin for the probe. This work was supported in part by grants from The Wellcome Trust and Action Research for the Crippled Child.

References: 1) Royer-Pokora *et al.* (1986) *Nature* **322**, 32–38. 2) Battat and Francke (1989) *Nucl. Acids Res.* **17**, 3619.

D7S449 detects a HindIII polymorphism tightly linked to the MET gene on chromosome 7

Michael Dean*, Claudia Stewart, Anjanette Perry, Dora Stauffer¹, Brith Otterud¹, Ray White¹ and Mark Leppert¹
Program Resources Inc./DynCorp, Frederick Cancer Research and Development Center, Frederick, MD 21702 and ¹Howard Hughes Medical Institute and Department of Human Genetics, University of Utah, Salt Lake City, UT 84132, USA

Source/Description: IEF 49 contains a 8.4 kb PstI fragment isolated from the IEF hybrid cell line (1). The insert is cloned into pBR322.

Polymorphism: HindIII detects a single two-allele polymorphism:
Allele A1: 4.4 kb Allele A2: 3.3 kb

Frequency: Estimated from 168 unrelated Caucasians
HindIII 4.4 kb allele: 0.87
 3.3 kb allele: 0.13

Not Polymorphic For: AvaII, BamHI, BglII, EcoRI, MspI, PstI, SacI, TaqI.

Chromosomal Location: Isolated from a hybrid containing only 7cen and 7q3 sequences. Tightly linked to the Met gene, on 7q31 (2) by multipoint analysis in CEPH pedigrees.

Mendelian Inheritance: Co-dominant segregation of the HindIII alleles observed in 56 families (660 individuals).

Probe Availability: Freely available from M.D.

Other Comments: RFLPs were observed under normal hybridization stringency; the probe is single copy.

Acknowledgements: This project has been funded at least in part with federal funds from the Department of Health and Human Services under contract N01-CO-74102 with Program Resources, Inc. By acceptance of this article, the publisher of recipient acknowledges the right of the U.S. Government to retain a nonexclusive, royalty-free license in and to any copyright of the article.

References: 1) Arfin, S.M., Cirullo, R.E., Arredondo-Vega, F.X., and Smith, M. (1983) *Somatic Cell Genet.* (9), 517–531. 2) Dean, M., Park, M., Le Beau, M.M., Robins, T.S., Diaz, M.O., Rowlet, J.D., Blair, D.G. and Vande Woude, G.F. (1985) *Nature* **318**, 385–388.

Editorial footnote Since this manuscript was submitted and accepted by my predecessor, another manuscript describing the same polymorphism was accepted and published (*Nucleic Acids Res.*, **18**, p. 4966) confirming the results.

* To whom correspondence should be addressed