Xba I polymorphisms in the 5' region of human erythrocyte alpha spectrin gene (Spna)

L.Kotula and P.Curtis

The Wistar Institute, Philadelphia, PA 19104, USA

SOURCE AND DESCRIPTION OF CLONE: 1.4 kb Xba I-Sal I genomic fragment located approximately 28.0 kb away from the 5' end of the human erythrocyte alpha spectrin gene subcloned in pUC18.

POLYMORPHISMS: Xba I digestion on total human DNA reveals two allele 3.5 kb and 3.0 kb polymorphisms.

ALLELE FREQUENCY: Determined by studying 37 unrelated individuals:
3.5 kb allele: 0.60
3.0 kb allele: 0.40

40.5% of studied individuals were heterozygotes.

NOT POLYMORPHIC FOR: Hinc II , EcoR I , Msp I , Pvu II , Sac I and Dde I.

CHROMOSOMAL LOCALISATION: Alpha spectrin gene has been mapped to chromosome 1 by hybridisation to somatic cell hybrids and localized to 1q22-1q25 by in situ hybridisation (1).

MENDELIAN INHERITANCE: Codominant segregation observed in 3 families (4 informative meioses).

PROBE AVAILABILITY: Request for probe should be sent to Dr Peter Curtis at the above address.

REFERENCE: 1. Huebner et al., PNAS (1985) 82: 3790-3793.

ACKNOWLEDGMENTS: This work was supported by NIH grant No. HL33884. Many thanks for providing either blots or DNA samples to L. and M. Showe, A. Linnenbach and J. Kant.