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Bgl II RFLP recognized by a human IRBP cDNA localized to chromosome 10

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SOURCE/DESCRIPTION: A 2184 bp cDNA (H·4 IRBP) encoding human interstitial retinol-binding protein (IRBP; Fong et al., 1984) isolated from a human retina cDNA library in λgt10 by screening with a bovine IRBP cDNA probe (B·23; Liou et al., 1986).

POLYMORPHISM: Bgl II (AGATCT) identifies a 2-allele polymorphism with bands at 6.3 kb (A_1) and 4.3 kb (A_2). There is a strong invariant band at 2.5 kb.

FREQUENCY: In 43 unrelated individuals (41 Caucasians, 2 Blacks)

A_1	6.3 kb	0.85
A_2	4.3 kb	0.15

NOT POLYMORPHIC FOR: Hind III, Pvu II, Bcl I

CHROMOSOMAL LOCALIZATION: Mapped to chromosome 10 using somatic cell hybrids. In situ hybridization suggests regional assignment near p11.2 → q11.2 with a secondary site of hybridization at q24 → 25.

MENDELIAN INHERITANCE: Demonstrated in two 6-member families (see Fig.).

PROBE AVAILABILITY: Available to collaborators.

REFERENCES: Fong, S.-L. et al., J. Neurochem., 42:1667-1676 (1984).
Liou, G.I. et al., Vision Research 26:1645-1654 (1986).

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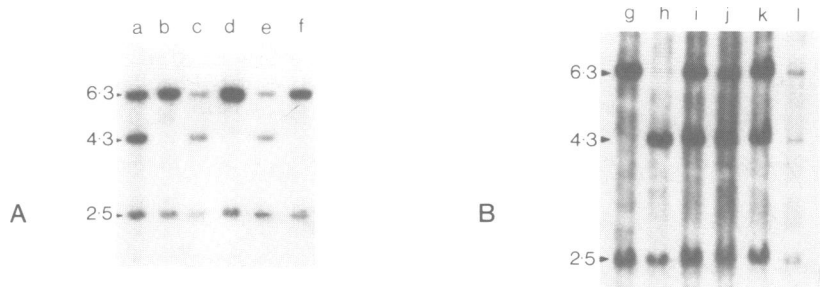

Southern blot analysis of Bgl II digests from two 6-member families (A, 0.8% agarose, 6 μg DNA; B, 0.7% agarose, 7 μg DNA). Fathers, lanes a & g; mothers, lanes b & h; offspring, lanes c-f, i-l. Gels calibrated with Hind III-digested λDNA.