

X mapping in man: evidence against direct measurable linkage between ocular albinism and deutan colour blindness

Summary. A Newfoundland kindred in which ocular albinism and deutan colour blindness are segregating provides strong evidence against the loci for these two X-borne characters being within direct measurable distance of each other.

The locus for ocular albinism is about 15 cm from *Xg* (Fialkow, Giblett, and Motulsky, 1967; Pearce, Sanger, and Race, 1968; Hoefnagel, Allen, and Walker, 1969; Pearce, Johnson, and Sanger, 1971), and *Xg* is not within direct measurable distance of the *deutan*, *protan*, *G6PD*, *haemophilia VIII* cluster of loci (Race and Sanger, 1975). It could, therefore, be assumed that *ocular albinism* and *deutan* are unlikely to be within measurable distance of each other, and the family here reported goes a long way to prove this assumption correct.

The family in the Fig. is part of a very large Newfoundland kindred in which ocular albinism is segregating. The ophthalmological details and history of this remarkable kindred which included 29 affected males and 50 heterozygous females were recorded by Johnson, Gillan, and Pearce (1971): it contributed to the demonstration of the *Xg* linkage (Pearce *et al.*, 1971).

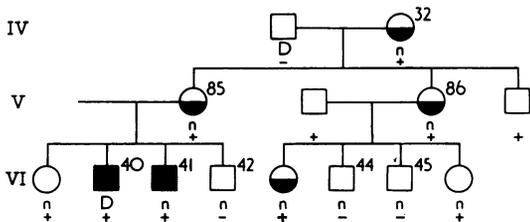


FIG. That part of the Newfoundland kindred which gives the evidence for lack of direct measurable linkage between ocular albinism and deutan colour blindness. Black = ocular albinism, half black = heterozygous ocular albinism (detectable ophthalmologically), white = normal. D = deutan, n = normal colour vision. + = *Xg(a+)*, - = *Xg(a-)*.

In a more recent visit to Newfoundland a section of the kindred was found to give the first direct information about the linkage relation of ocular albinism to deutan colour vision.

The colour vision tests used were (1) American Optical Hardy-Rand-Rittler (H-R-R-) Pseudoisochromatic Plates, (2) Ishihara Pseudoisochromatic Plates (1970), and (3) Farnsworth Panel D-15 Dichromatous Test. The answers are shown in the pedigree (Fig.), where the generation and individual numbers are those of Johnson *et al.* Between *ocular albinism* and *deutan* there are 4 recombinants (VI. 40, 42, 44, 45) to 1 non-recombinant (VI. 41) for which the lod scores are -2.541 at $\theta=0.1$ and -0.742 at $\theta=0.3$, good evidence against the detectability of linkage.

During this recent visit to Newfoundland blood was taken from one member of the family (VI. 45), not available in 1971 and who provided one more non-recombinant between *ocular albinism* and *Xg*. His addition to the total account (Pearce *et al.*, 1971) lowers slightly the best estimate of the distance between the loci to 14.3 cM with probability limits of 0.09 and 0.25.

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