A Color Vision Anomaly Showing Holandric (Y-linked) Transmission

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ALL previously published pedigrees of red-green color blindness or color deficiency seem to show sex-linkage and appear to be typical recessive, or nearly recessive, traits due to genes carried on the X chromosome. The defect represented in the pedigree to be presented here does not behave in the usual manner but is apparently a case of holandric or Y-borne heredity.

Ichthyosis hystrix gravior, hypertrichosis of the ears, and a type of webbed toes are examples of characters dependent upon genes assumed to be transmitted via the non-homologous part of the Y chromosome. Such holandric genes are assumed to be passed on to future generations solely through the male line from father to son. The females never exhibit an holandric character nor transmit it to their progeny. It is not possible to state whether holandric characters show dominance or recessiveness, due to the lack of any allele corresponding to the gene of interest.

In the pedigree of the family presented in fig. 1, it is reasonable to assume that the color-anomalous condition is transmitted only through the male line. The females neither exhibit nor carry the trait according to this assumption.

Unfortunately, it has not been possible to test the members of the family in a satisfactory manner. Testing this somewhat scattered family with the anomaloscope was out of the question. None of the females were tested in any way. In the pedigree, *II-9*, *II-14* and *III-7* were tested by the pseudo-isochromatic plates of the American Optical Company and found to be color deficient. The individual *III-1* was refused admission to the Army Air Corps, World War II, because of his color-discrimination deficiency. The propositus (R. K. Cambier, *III-4* of the pedigree) was tested at the Ophthalmology Clinic of the University of Minnesota. The yarn test did not reveal positive data but this test is thought to be less critical. The Stilling-Ishihara pseudo-isochromatic plate tests showed a definite color deficiency. Fifteen of 46 items were missed by both eyes.

While the testing of the members of the family was inadequate, the written or verbal statements were very definite. Each person was quite certain as to his or her possession of, or lack of, a color vision anomaly. The correlation between being a male descendant of *I-1* and having the color anomaly had been recognized in the family for many years.

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If the evidence presented by the members of this family is accurate, it is highly unlikely that the heredity of this color vision anomaly is of the usual X-linked type. We have calculated the probability that this pedigree, as shown, represents the X-linked type of heredity and the probability is negligible.[1]

An alternative assumption is that we have a case of the usual X-linked gene for color deficiency having crossed over to the non-pairing part of the Y chromosome due to some special circumstances of an unusual nature. Were this the case we might expect that the normal allele of the color deficiency gene would still prevent expression of the anomalous gene in the new location in males. This alternative explanation also seems to be an unlikely one.

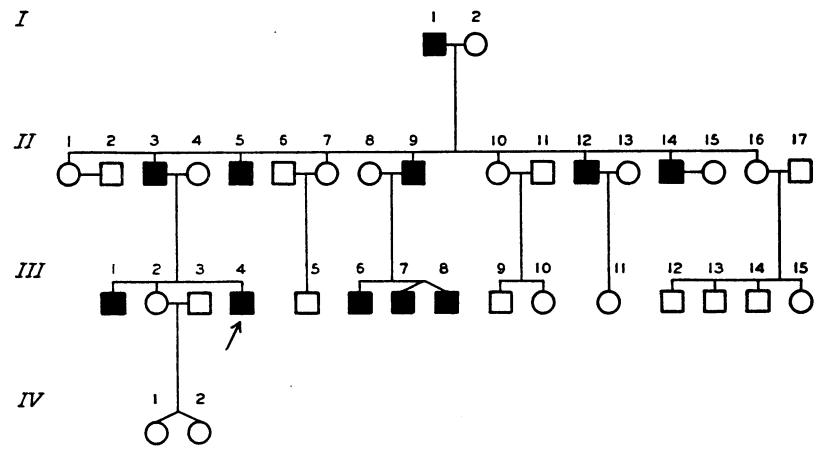

FIG. 1. Pedigree of the Cambier family. The inheritance of the color vision anomaly through the male line is shown by the black squares representing the affected males. The arrow indicates the propositus.

It is most logical to assume that we are concerned with a distinct mutation in the non-homologous part of the Y and that this mutation has no allele on the X chromosome. The Y-borne mutation is therefore an holandric gene with similar effects to the usual X-borne genes but is probably somewhat different in the details of its action.

DISCUSSION

Gates (1946) gives an extensive review of the literature on the inheritance of color-blindness. From his account it seems that there may be two loci on

[1] Given the presence of colorblindness in the propositus and given the sex distribution shown in the pedigree, the probability (P) of finding the reported distribution of color-blind and normal relatives is, on the hypothesis of recessive X-linked inheritance, $P = (\frac{1}{2})^{14}q^2$. Here q stands for the frequency of all X-linked alleles causing color-blindness in males but not in heterozygous females; if we assume $q = 0.1$ then $P = 1/1{,}638{,}000$. (In this calculation it is assumed that *III-7* and *III-8* are dizygotic twins.)

the X chromosome concerned with red-green color-blindness of the usual sort. One locus is concerned with deuteranopia, or green-blindness, and the other with protanopia or red-blindness. Gates also reproduces Cunier's famous pedigree in which color blindness is inherited from mother to daughter only, through five generations. This is a case of hologynic inheritance, the opposite of the holandric type of heredity shown in the Cambier pedigree.

In addition to the two loci for red-green color-blindness on the non-homologous part of the X chromosome and a third locus on the non-homologous part of the Y, shown in the Cambier pedigree, there is a fourth locus concerned with achromatopsia which Haldane (1936) considers to be located in the homologous part of the X and Y chromosomes.

We wonder if it is purely coincidental that all four loci known so far, which are concerned with color vision, are on the sex chromosomes and none on the autosomes. It would seem likely that more than coincidence is involved, though we have no explanation at hand which has experimental evidence to substantiate it.

SUMMARY

A gene causing a deficiency in color vision has been shown to be inherited in the holandric fashion, that is, always through the paternal line. Females could never show this type of color vision deficiency nor carry the gene for it, as the gene seems to be located in the non-pairing section of the Y chromosome.

It is assumed that the gene for color vision deficiency in this family represents a mutation in the non-homologous section of the Y chromosome and that it has no allele in the X chromosome. It has similar effects to those of the usual X-borne genes but probably differs in the details of its action.

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