HAEMOPHILIA AND COLOUR BLINDNESS OCCURRING IN THE SAME FAMILY

BY

W. J. B. RIDDELL

GLASGOW

The association of haemophilia and colour blindness in one family is not common. The family about to be described was found as a result of deliberate search amongst known haemophilic families in the West of Scotland. Seven haemophilic pedigrees were investigated as far as practicable and the affected members were examined for colour defects. Ishihara’s iso-chromatic plates were used in all cases and the findings were confirmed by another method which varied according to circumstances. Stilling’s plates, the Edridge-Green lantern, bead, and wool tests were used as confirmatory tests. Introduction to the families was obtained through hospital and clinic records, where the diagnosis of haemophilia had been definitely confirmed in at least one member of the family. I am very much indebted to the physicians and surgeons who have permitted me to examine their patients and have access to their records. Only one pedigree was obtained from a dental clinic which was undoubted haemophilia. Prolonged haemorrhage following the extraction of teeth, without other clinical evidence, and no definite family history, is not uncommon in both male and female children. Eight children of this type were examined and no colour defects were found. Such cases are not true haemophilia.
The relevant portion of the pedigree is shown in the diagram.

I. 1. Mrs. H., aged 52 years (1936), is a healthy working-class woman of small stature. She is the fourth child of a family of seven, three males and four females. Three brothers and two sisters are married and have families. None of them is a bleeder. Her father died when seventy and her mother when eighty-one. She has no knowledge of bleeders amongst her relatives. Her colour vision is normal.

II. 1. T. H., aged 33 years (1936), is an unemployed hammer-man. He is a healthy looking man and has had no serious illness.

He is not a bleeder. In August, 1936, he was examined with Ishihara’s cards with the following result. 12.5.6.2.21.---.---5.2.---.---. This indicates red-green blindness. Unfortunately this man would not agree to more detailed examination. Several attempts were made to get his co-operation without success. When examined in August he informed me that he had no difficulty in reading coloured numbers, which he had seen before when examined for service purposes. This point was inquired into through official channels and the following information was obtained. He was examined in July, 1930, with Ishihara’s test with the following result. 12.3.5.2.21.---.---.5.2.---.---. With the lamp, green after signal green was called white. Yellow after red was called white. Both with a number three aperture. His central vision and fundi were normal.

This man is not a bleeder, but is a colour defective person. He is married and has one daughter aged 7 years—III. 1.
II. 2. This woman is a married daughter, aged 29 years (1936). She lives in Canada and has one daughter—III. 2.

II. 3. This boy was a bleeder and died when four years old.

II. 4. This boy was a bleeder and died when three months old.

II. 5. J. H. was also a bleeder and was brought up with great difficulty owing to frequent disabling haemorrhages. He died aged 21 years. Photographs of this member of the family showed a well-developed healthy looking youth.

II. 6. H. M. is a healthy girl of 18 years. She has never shown any tendency to bruising or of bleedings into joints.

II. 7. R. H. is 12 years old (1936). He has bruised easily and has had swellings of his joints ever since he was born. On one occasion he lost blood for a fortnight following upon a tooth extraction. He has been in hospital on two occasions and has required blood transfusion. Two years ago he was struck on the left wrist by a cricket ball. This injury was followed by haemorrhage into the joints and anaesthesia of the fingers. He subsequently burned his fingers. The tips of three of them became gangrenous and were lost. He is a well-nourished and healthy looking boy and is normally developed for his age. His education has been handicapped severely owing to his frequent absence from school.

In August, 1936, he was examined with Ishihara’s cards with the following result. 12--2. 21--5. 2--. This test was repeated in October, 1936, when the answers given were the same. The defect was confirmed by means of Stillings’s plates (nineteenth edition, 1936, Leipzig). In this series he read Nos. 1, 2, 7, 9, 24 and 31 correctly. In Plates 16, 18, 20, 32 and 33 he read one figure correctly and the other incorrectly (these plates consist of two numerals). He was unable to see any figures on Plates 3, 4, 5, 6, 8, 10, 11, 12, 13, 14, 15, 17, 19, 21, 22, 23 and 34. Plates 25 to 30 inclusive were not used in the test. The test was repeated twice, in good daylight. His central vision and fundi were normal.

This boy is a bleeder and is colour defective.

Haemophilia and colour blindness are both sex-linked characteristics. In this family there are two surviving males. Both have colour defects and one is a haemophilic. Three other male children were haemophils and died young. The two daughters have always been healthy. The mother (I. 1.) came from the North of Ireland and has lost touch with some of her kinsfolk. No history of haemophilia could be obtained in preceding generations. The genetic significance of this family is of great interest as may be seen in a forthcoming paper by Julia Bell and J. B. S. Haldane, on “Linkage of haemophilia and colour blindness,” where it is discussed with other illustrative examples; it is unfortunate that more details are not available.
Throughout the investigation of the seven haemophilic pedigrees the possibility of congenital or developmental abnormalities in the eyes of members of these families was kept in mind. No such defects were observed with the single exception of the family above described. In conclusion I must express my very deep gratitude to Miss Julia Bell for her encouragement and assistance in the study of the non-haemophilic pedigrees and in the preparation of this note.

"TWINCENTRIC" LENSES

BY

F. A. WILLIAMSON-NOBLE

LONDON

In the August, 1936, number of this journal, a short article was published describing a type of bifocal lens in which the optical centres of the distance and reading portions were made to coincide, thus eliminating any vertical "jump" when the direction of gaze was shifted from the upper to the lower portion of the lens. The same effect is produced even if the centres do not coincide, provided they are on the same horizontal level. It is therefore possible to make a bifocal lens with the centre of the reading portion displaced inwards to the correct amount without producing "jump."

There are two practical methods of obtaining this result. The first is applicable to a solid bifocal and consists in grinding the lower segment so that its optical centre is on the same level as that of the distance portion. This works quite well, but has the disadvantage of producing a conspicuous reading segment, the upper
HAEMOPHILIA AND COLOUR BLINDNESS OCCURRING IN THE SAME FAMILY

W. J. B. Riddell

doi: 10.1136/bjo.21.3.113

Updated information and services can be found at:
http://bjo.bmj.com/content/21/3/113.citation

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/