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COMMUNICATIONS

COLOUR BLIND FEMALES

THE INHERITANCE OF COLOUR BLINDNESS IN
MAN (*concluded*)

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AMONGST the comprehensive literature concerning colour blindness the works of Dalton, Horner, and Nettleship, are some of the most valuable. Even before the time of Dalton, however, the hereditary character of the anomaly was touched upon, so that one nearly agrees with Magnus's statement in 1877: "The hereditariness of colour blindness has been known as long as the defect itself. All who have seen many colour blind, will have often noticed that it was in their mother's family that other cases were found." The occurrence in families was already mentioned in the very first case of colour blindness described (Joseph Priestley in 1777), from which we learn that the shoemaker, Harris, had two colour blind brothers. When describing his own anomaly, Dalton wrote also of his two colour blind brothers, mentioning a total number in his work of 40 colour blind men (1798).

In his small but very important treatise, "Experiments on Colour," Lord Rayleigh first describes his apparatus by means of which he can obtain comparatively pure colours, in a manner enabling him to compare a pure yellow colour with another yellow colour obtained by a mixture of red and green (Rayleigh's equation). Thereafter he states that a red-green mixture, which to himself and most beholders appeared like a pure yellow, corresponding to the D line in the spectrum, appeared to his three brothers-in-law Balfour as red, "almost as red as sealing wax." In order to suit the colour mixture to their eyes the red part had to be made so weak that the mixture appeared definitely green to normal eyes. The three brothers-in-law agreed mutually in their colour perception, and they all accepted the identical equation (the deuteranomalous equation). A fourth brother-in-law and three sisters-in-law had normal colour perception (1881).

FIG. I.

Family Scott.

We possess from the 19th century and the last twenty years of the 18th century a number of papers and pedigrees referring to colour blindness. It will be shown below how the anomaly in these tables, perfectly and without exception, falls into line with the interpretation of *colour blindness as a recessive sex-linked character*. This aim has necessitated a careful examination of the original pedigrees, as faulty and incorrect extracts from some of them have been quoted again and again in text-books and periodicals as exceptions from the rule, which in reality is unbroken.

The first, and even now one of the best pedigrees, was published by Lort in 1778, based on a letter from Scott, in which he describes his own condition and that of his near relatives. Scott states:

"My business was behind a counter many years, when I had to do with variety of colours. I often, when alone, met with difficulty, but I commonly had a servant in my way to attend me, who made up my deficiency. I have been now several years from trade. My eyes, thank God, are very good at discerning men and things." And further: "My mother's one brother had the like impediment with me, though my mother knew all colours very well."

In spite of the few persons included in this pedigree, it really shows most of the facts needed; hence it will be given here.

We must assume that Scott had inherited the anomaly from his mother, who most probably was a conductor, as she had a colour blind brother and got a colour blind son.

Thus the fact that one of Scott's sisters was colour blind is explained naturally enough, as she may have inherited the defect both from her father and her mother. The sons of this colour blind sister were all as a matter of course, colour blind, while Scott's own children were colour efficient. All these facts are now naturally and easily explained by the laws.

Scott's sister is the first colour blind woman ever described. It is true, that as early as 1683, Tuberville published his extraordinary case: "A maid, 22 years old, came to me from Banbury who could see very well, but no colour besides black and white. She had such scintillations by night (with the appearances of bulls, bears, etc.) as terrified her very much. She could see to read sometimes in the greatest darkness for almost a quarter of an hour."

Usher seems to be correct when saying, with regard to this case that "it was probably total colour blindness with day blindness or possibly some hysterical condition."

The brief papers of Harvey, J. Butters, Nicoll, Sommer, and Rozier, may just only be mentioned in passing on to the case of James Milne, published in 1825 by Combe, who describes colour blindness in J. Milne, his two brothers, his mother's father, and one male cousin. This good pedigree was continued by Usher in 1912, showing how the anomaly adheres to the typical rule throughout seven generations.

The hitherto most inexplicable pedigree of colour blindness was published in 1838 by F. Cunier, who himself admits that it varies in a very marked degree from all similar tables published previously. In this case the anomaly is supposed to have been passed on from mother to daughter in five successive generations, attacking all females (12) while all males (8) went free. Cunier states, that while examining a married lady of 58, he learnt that she was colour

blind, inasmuch as she never saw any difference between dark blue and cherry red colours. He was allowed personally to test her daughters and her daughter's daughters by means of skeins from a work basket. Further, he learnt that the maternal grandmother, mother, and the only aunt of his patient had suffered from the same inability, while all male members of the family had a good colour sense.

The anomaly described by Cunier cannot, however, have been the hereditary red-green blindness, for a closer study of his original paper reveals the following description of the pathological findings in their eyes, as most pronounced in the eldest daughter and daughter's daughter of his patient. "The pupil has a normal shape and reacts normally, but has not the usual black colour; in the depths of the eye (*'au fond de l'oeil'*), slightly above the horizontal meridian and outwards, an oval yellowish spot is seen, with a green dent in the centre." These facts he interprets as indicating *'l'absence du pigment noir de la chorioïde'*.

"These women did not endure bright sunlight, especially when coming out in the sun from a dark room, or when the sun suddenly broke through a cloud."

The male members of the family had quite normal eyes, and showed neither colour blindness nor photophobia. It was stated that no disease of the eyes nor any case of blindness was known in the family, and this fact Cunier thought remarkable, because until that time he had found defective sense of colour only "*dans l'amaurose ou dans la cataracte'*" Cunier naturally did not possess any ophthalmoscope and was therefore not capable of making any exact diagnosis. We are, however, justified in assuming the alterations in the eyes to be the cause both of photophobia and imperfect sight as well as of their defective colour sense. Herbert Parsons is undoubtedly right when stating in his "Pathology of the Eye": "Cunier published a remarkable family, in which through five generations only women were affected, but the case is almost certainly not one of congenital colour blindness."

In 1840, Szokalski's paper on colour sense was published, containing a chapter on colour blindness, giving, however, no fresh pedigrees. Concerning Cunier's information he says: "*une observation très curieuse, qui est dans une contradiction flagrante avec toutes celles qui ont été recueillies, jusqu'à ce moment.'*"

Dr. Pliny Earle of Philadelphia in 1845 described his own colour weakness, adding a very comprehensive pedigree, that is unusually clear: Out of 3 colour blind brothers 1 has 5 normal daughters, who together have 11 colour blind sons, and 1 normal daughter's daughter, who has 1 colour blind son. The pedigree has been carried forward in an excellent manner by Nettleship and Usher and was published in 1915.

Holmgren rendered great services by his extensive research work concerning the occurrence of colour blindness in Sweden, and he also was deeply interested in the problem of its hereditary nature, which he has dealt with in several publications. He has, however, never published any pedigrees.

The usual hereditary type of colour blindness was first demonstrated and described by the Swiss ophthalmologist Horner, later affirmed by Burckhardt and Bollinger. Horner in his very significant work "Hereditariness of Daltonism," gives 2 uncommonly fine pedigrees of red-green blindness in 6 and 4 generations respectively, and on the basis of these two he formulated his well known law, viz.: "that sons of daughters whose fathers were colour blind are most likely to be the same, although not without exception; or colour blindness is transmitted in the reversible type from grandfather to grandchild."

In most references to Schoeler's instructive pedigree there occurs an inexactitude, which causes it to appear inexplicable by the laws concerning a recessive sex-linked character, although the pedigree *per se* is unusually fine and typical.

The original table of Schoeler is this:

FIG. II.
Schoeler's pedigree.

A married couple had 5 sons, all colour blind, which fact might indicate that the mother was colour blind, only no statement is made in this respect. One of these sons had 3 daughters, of which 2 were colour blind, which proves that their mother must have been a conductor. And 1 of the 2 colour blind sisters (in a marriage with her colour efficient cousin) had only colour blind sons and two normal daughters.

In the inexact quotation* of this table the six sisters and brothers of the fourth generation, are wrongly said to be children of No. II, with no fourth generation and the occurrence of the anomaly remains quite inexplicable.

Faulty explanations of the pedigrees showing colour blindness are often brought forth by various authors, who thus procure examples of anomalies that seemingly do not fall in with the laws. Thus, several authors have mentioned a pedigree published by Stilling explaining the colour blindness of two brothers as an inheritance from the grandfather through the unaffected father, while the anomaly is explained in a much more natural way as inherited from the mother (a conductor), the wife of the unaffected father.

Further, Sir William Abney in his book on colour vision mentions several cases where, in his opinion, the anomaly has been transferred from father to sons, and he even supposes Dalton's father to have been colour blind. Even if that was the case, we should conclude from what has been said above, that Dalton did not inherit his anomaly from the father, but from the mother, who was probably a conductor.

A series of investigations undertaken by Cohn in 1878 shows very clearly that when colour blind men have colour blind sons the fact does not imply any cause and effect. "He found among his one hundred colour blind, three times the father red blind, in fourteen cases the brothers colour blind, and once the father and three sons. In no case were all the children colour blind." As found with the incomplete tests of the time the percentage of colour blindness being just about four, no better proof can be desired that the colour blindness of the fathers has no influence on the colour perception of their sons. In the same work Cohn has a small pedigree: A colour blind man had a sister, whose sons were all colour blind; his mother's brother had a colour blind son and a colour blind grandson through his daughter. Even if the mother's brother were colour blind himself, of which we have no information, his son, at all events, must have inherited the anomaly from the mother.

As is well known, Albrecht Nagel was red-green blind, and he has himself described his anomaly. His pedigree shows that a colour blind father and a colour efficient mother (who must have been a conductor) had two sons of whom one was colour blind, and three daughters of whom two were colour blind. This pedigree is referred to in L. Plate's great text-book as the only known example of red-green blindness occurring as a dominant character, and Plate adds: "The rule, as is well known, being that the red-green blindness is a dominant sex-limited anomaly (dominant in ♂ recessive in ♀)!"

R. C. Punnet makes partial use of the same terms in his book,

* Nagel's *Jahresbericht*, 1878, p. 117.

"Mendelism;" but in these terms we cannot agree. Also he states, that "since neither colour blind men nor heterozygous women (conductors) are common, the chances of a marriage between them are very rare." Above it has, however, been proved that there is every reason to believe that such marriages are not so rare—1·8 per cent.

The best investigations concerning the hereditary character of eye-defects and anomalies were those carried out by E. Nettleship, the late lamented ophthalmologist. Most of his pedigrees deal with the hereditary eye-defects and are found in his admirable Bowman lecture, "On some Hereditary Diseases of the Eye";* as, however, hereditary colour blindness must be considered an anomaly

FIG. III.

Rivers's pedigree from the Todas.

more than a disease, he has not dealt with this subject in the work cited. All the same, his industry produced 10 pedigrees of colour blindness,† of which about half are almost completely worked out. After his death, C. H. Usher arranged and published further four pedigrees, of which one is a continuation of Pliny Earle's excellent pedigree from 1845.

Nettleship, also, was much interested in colour blindness in women, and he succeeded in examining or obtaining information of about 20

* *Trans. of the Ophthal. Soc.*, Vol. XXIX.

† *Trans. of the Ophthal. Soc.*, Vols. XXII, XXIII, XXVI, XXVIII, XXXII.

such cases. Three times he found the anomaly in one of twins. Twice the twins were girls, and at any rate one couple was proved to be mono-oval. They were as like as two drops of water; but one was colour blind, and remarkably enough, the other was found to be colour efficient. They were not, however, more than $8\frac{1}{2}$ years old at the last examination. (A further examination with our modern tests of these twins and of their issue would be of very great interest.) Once the twins were of different sex, and, of course, the boy was the one colour blind.

As a curiosity, I give below a pedigree from W. H. R. Rivers's investigations among the Todas mentioned above, where the anomaly occurs in two families related to each other, but only in one of them the defect can be imagined as coming from the common origin.

It is necessary to mention Dr. G. F. Göthlin's work, he being the first to elaborate amply detailed genealogical tables based upon personal examinations of all members of his families, who are at present alive. He has employed the best colour sense tests in existence in our time, and, what is more important, he has as far as possible *invariably* used spectral colour mixtures, partly by means of Nagel's anomaloscope, partly by his own portable polarisation anomaloscope. His work, therefore, presents a more scientific aspect, giving a greater value to his results than all the previous investigations that were partly based on descriptions by word of mouth and traditions, partly on tests carried out with inferior testing apparatus. It must be admitted, however, that Nettleship also to some extent made use of spectral colour mixtures, and, as above quoted, he has brought forth many very admirable pedigrees. Göthlin's lecture in the Psychological Research Institute in Upsala, in December, 1915, was a preliminary summary of the results of four years' work, and interesting things may be expected from his further publications. Amongst a number of families he has selected eight, whose pedigrees he publishes in his treatise; the pedigrees are not very extensive, but so good and so fully detailed that he must be considered justified in drawing his important conclusions.

Thus, for instance, he appears to have proved Holmgren's and Abney's supposition, viz.: that "the nature of the colour blindness within the same family is always the same," to be not always reliable. On the basis of the previous statement, we cannot, however, agree with Göthlin, when he concludes: "Personally, I am of the opinion that when colour blindness is very marked in the father, the inheritance may be manifest even in women who are heterozygots." He believes that he has seen two such cases, but in both the family of the mother has hardly been examined; it is true that in one case the examination included the mother's

brother, sister, and sister's daughter, but that is insufficient to prove anything. Göthlin finishes his conclusions as follows :

"The result of this research among families proves that there exists between a number of colour sense types a genetic connection which has not previously been demonstrated. A genetic connection has thus been discovered between protanopia and deuteranopia, and also between deuteranopia and deuteranomalialia, probably also between protanopia and protanomalialia." In his paper he goes into the problem more deeply, expressing partly the same views which Köllner just as strikingly has set forth and which, if they prove to be correct, will greatly simplify our conception of the various forms of anomalies of colour sense. Köllner states : "The Rayleigh equation of the deuteranomalous may be changed into the equation of the protanomalous by the simple process of imitating, by means of a suitable absorbing medium, the conditions of light under which the protanopes and the protanomalous distinguish themselves from the deuteranopes and the deuteranomalous. In spite of the varying Rayleigh equations we may therefore suppose that the three colours in question (red, green, yellow) may have approximately the same colour value for the protanomalous as for the deuteranomalous. Further, we may imagine that the difference between the Rayleigh equations of the two forms may be dependent in the first place on varying values of light."

In my own investigations of colour blind families, I have especially aimed at finding the highest possible number of red-green blind women, because in their families one would presumably find numerous red-green blind persons. Experience has proved the assumption to be right, since precisely by seeking colour blind women I have found families most decidedly suited for further investigations.

In the course of the last two years and aided by kind colleagues* I have had the opportunity of examining very closely 16 colour blind women (dichromatics and anomalous trichromatics), their families, and nearest relations. Together with the above-mentioned 20 school girls, this will make 36 cases of colour blind women. Later on I hope to publish some of my pedigrees, which *all* show how the anomaly is inherited in the typical manner ; whenever I have been able to ascertain the facts, these colour blind women had colour blind fathers and colour blind sons.

Conclusion : The results obtained by a search through medical literature and through all pedigrees published with regard to red-green blindness as an hereditary character, will thus be seen to conform completely with the results of my own investigations, proving that the congenital red-green blindness invariably and

*I beg to express my special gratitude to the ophthalmologist Dr. Harald Gjessing, Drammen.

without exception follows the laws of inheritance as a *recessive sex-linked character*. The few pedigrees which have previously been quoted as exceptions from the ruling principles, have on a closer examination of the original papers proved to be partly misquoted and partly to be concerned with anomalies other than the congenital red-green blindness. No single exception can be demonstrated to the unyielding principles.

It is true, that Bateson in his book, "Mendel's Principles of Heredity," 1913, mentions quite casually a case of an alleged colour blind mother with an alleged colour efficient son. The case has been brought to his notice by Miss J. E. Downey, of Wyoming University, but it cannot be fully accepted until all details of the case have been investigated. Among other things it must be excluded that the mother's colour blindness is acquired; also it is necessary to know what sort of colour tests were employed.

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DR. EDRIDGE-GREEN'S THEORIES OF VISION

(concluded)

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VIII. The Evolution of the Colour Sense

Dr. Edridge-Green's views on the Evolution of the Colour Sense are based on the following considerations:—(1) Psycho-physical theory; (2) Colour vision in primitive races (*Trans. Ophth. Soc.*, Vol. XXI, 1901, p. 182); (3) The order of disappearance of colours in the spectrum as the intensity is gradually diminished (Edridge-Green's "Physiology of Vision," p. 169); (4) The atavistic theory of colour blindness.

(1) *The Psycho-physical Theory*.—"The sense of sight must have been first developed for those waves which produced their maximum effect upon the sensitive protoplasm" (p. 213). "The wave-length of the physical stimulus is the physical basis of the sensation of colour" (p. 214). "The eye would first discriminate between the rays which are physically most different—the red and violet, that is presuming that the eye had become sensitive to this range" (p. 214). In this stage "the luminosity curve should be the same as the normal sighted" (p. 216). "The next stage in the evolution of the colour sense was when a third colour appeared at the third point of physiological difference, that is, in the centre of the spectrum in the position of the green" (p. 216). "As evolution proceeded the power of differentiation occurred in the regions between the red and the green and the green and the violet until a stage was reached in which a fourth colour, yellow, was seen at the next point of greatest physiological difference" (p. 217). "The next step in the process of evolution occurred when the retino-cerebral apparatus was able to differentiate a fresh colour between