On cases of congenital day-blindness with colour-blindness / by Edward Nettleship.

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ON CASES

OF

CONGENITAL DAY-BLINDNESS WITH COLOUR-BLINDNESS.

By EDWARD NETTLESHIP.

In the cases (about seventeen in number) here referred to, and eleven of which are detailed, one of the most prominent symptoms is, as the name implies, a defect of sight which is greater by day than in the evening or by artificial light. It is true that the patients do not usually come to us complaining of day-blindness, as the victims of retinitis pigmentosa come complaining of inability to see after dusk, it is for amblyopia sometimes with myopia that they apply. In addition to these symptoms there is always nystagmus, and very marked, sometimes complete, colour-blindness. The degree of amblyopia varies in different cases; in two cases (Nos. 6 and 7) it amounted to complete amaurosis. The defect is, so far as I have seen, stationary, and when the history is complete we always hear that the oscillation was noticed a few weeks, or, at most, about three months, after birth; the defect of sight may, therefore, be confidently dated either from birth or early infancy. The media are clear, and in many cases the fundus appears quite healthy; but in others (Cases 1, 3, and 7) slight changes are found, indicative of former neuritis or perhaps retinitis; it is to be particularly noted that there is never any pigmentation nor other indication of atrophy of the retina.[1]

[1] See Postscript.

In respect to cause I am only able to mention the significant fact that the disease appears often to occur in several children of the same parents, as pointing probably to inheritance of a morbid tendency, or possibly to the occurrence of actual intra-uterine disease of the optic nerves. Thus, my sixteen cases occurred in five families with thirty-five children.

Family I.—F—	. 11 children	. 6 cases.
„ II.—G—	. 5 „	. 1 „
„ III.—P—	. 6 „	. 4 „
„ IV.—C—	. 3 „	. 3 „
„ V.—G—	. 10 „	. 1 certainly,
		2 probably,
	—	—
	35	17

The theory of inheritance is strengthened by the occurrence of the disease in collateral branches of the same families (III and IV). The history of colour-blindness in an uncle of one of the patients (Case 1) may be noted in passing, though the account is too meagre to permit any conclusion as to whether it was anything more than ordinary congenital colour-blindness.

The association of the eye defect with feeble intellect, and even idiocy, is a point which these cases have in common with some other congenital or inherited affections of sight. As to the local condition we may conjecture, with a good deal of probability, that the symptoms depend upon a disease of the optic nerves behind the eyeball, a retro-bulbar neuritis, of which the intensity, and perhaps the precise seat, may differ in different cases, thus accounting for the exceptional occurrence in the worst cases of well-marked changes in the discs.

The peculiar symptom to which I desire to call special attention, the improvement of vision in dull light, is, in all probability, caused by the defect being greatest in the centre of the visual field, and thus forming a large and ill-defined central scotoma;[1] as, however, I was not able to measure the fields in any of the following cases, I can only conjecture that this explanation is correct. I may, however, point to the invariable

[1] For the explanation of this symptom *see* 'St. Thomas's Hospital Reports, vol. ix, p. 52.

association of central scotoma with improvement of vision in dull light, which we witness in tobacco amblyopia and in some other cases of acquired amblyopia, in support of this view; and to the view advocated by Leber, that a low degree of neuritis in the cortical layers of the optic nerve behind the eye furnishes the explanation of the symptoms in tobacco amblyopia and conditions resembling it.

I do not for a moment suppose that the cases narrated below are new to observers here or abroad, and have brought them forward simply as illustrations of one of the types of congenital amblyopia or amaurosis, which, so far as I can learn, has not yet received any separate notice. A case, apparently of the same kind, recorded by Donders in the 'Report of the Heidelberg Congress' for 1871, is referred to by Joye Jefferies in his work on 'Colour-Blindness,' p. 35, and Landolt ('Examination of the Eye,' p. 191) mentions four cases of congenital total colour-blindness with photophobia and pallor of the discs, which, not improbably, belong to the same category.

Family I. Case 1.—Miss Mary F—, æt. 25, tall, thin, very fair, but freckled skin, abundant light reddish-brown hair, teeth defective in enamel ("mercurial"), forehead at root of nose flattish. Hearing and other senses good; intelligent and excitable.

V. by daylight each $\frac{2}{2\,0\,0}$, and 6 J. at 4″; refraction E. or H. $\frac{1}{4\,0}$. Ps. active. Constant, slight, lateral nystagmus. Visual field (finger test) shows no contraction.

Her great complaints are that she cannot see so well by day as by artificial light, and that she cannot tell colours.

In respect to the former point she said that in the daytime her sight was so bad that she was afraid to cross the street, whilst at night she saw so well that she could do so with ease and comfort, and that she could read small print by a light so dull that other people had to put away their books.

Suspecting that this symptom was related to the size of the pupils I dropped eserine into the eyes, and found, as I expected, that as soon as the pupils were of pin-hole size, the sight had become much worse.

Of colours she has not the slightest appreciation; she sorts the wools entirely according to their brightness (luminosity);

all colours look to her like shades of black and white, and she always dresses in black and white to avoid making absurd mistakes. As a rule, shades of green look brighter than correspondingly dark shades of red, but the yellows are probably the brightest to her, for on looking through various coloured glasses she preferred a rather dark yellow, and said it improved her sight very much.

Atropine acted fully, and the sight was made much worse by it; probably a slight dilatation might have given some improvement.

Ophthalmoscope.—Disc and vessels quite healthy (erect and inverted); a whitish haze of doubtful meaning about the Y. S. No certain changes.

There is no history of fits nor other illness in infancy. Her sight has always been exactly as at present, and the nystagmus is said to have been noticed at the age of three weeks.

Case 2.—Miss Kate F—, æt. 20, the youngest sister (of the family of eleven), resembles her sister Mary in build and complexion and in character of teeth, but her hair is redder. The condition of her sight and eyes is exactly the same as in the former case, with the single exception that her nystagmus is much more marked. Like her sister's, the oscillation is said by the mother to have begun about three weeks after birth; she was healthy in infancy.

V. Each eye singly 8 J., together 6 J., at 3″ and $\frac{20}{200}$.

She behaves precisely like her sister with coloured wools and papers, being totally colour-blind, and her sight in daylight is similarly improved by looking through a yellow glass. She sees much better in the evening and by artificial light. The spectrum looks to her like a light band or stripe of one colour, brightest in the middle, and becoming darker at each end; it seemed from her replies to look darkest at the violet end. With coloured papers red always looked to her much darker than green of the same shade. Of Stilling's coloured letters on a black ground the *blue* was "much lighter" than the other colours (sometimes "white"), next the *yellow*, then *green*, which is "very dark," and lastly *red*, which is "nearly black."

The following is a list of the family to which the above two cases belong:

(1) Family I. F—, eleven children, of whom the six marked * are amblyopic and colour-blind.

The list of the children is as follows:

(1) Male, died of abscess of liver in India; was an army surgeon. Had perfect sight, fair complexion, and reddish hair.

(2) Male, died a baby of teething and water on the brain. Good sight.

*(3) Male, æt. 35. Colour-blindness and reported to suffer from defective sight in exactly same way as the two sisters whose cases have been narrated. Can see well enough to earn his living as a book-keeper. Reddish-brown hair.

(4) Female, died at the age of sixteen of "abscess of the leg" and "consumption." Sight perfect. Very fair skin and brown hair.

(5) Male, æt. 31. Sight perfect. Very fair skin and sandy-brown hair.

*(6) Female, died at the age of sixteen after a fall. Was colour-blind and amblyopic exactly like the other two sisters. Golden-brown hair.

*(7) Female, æt. 27. Believed to be colour-blind like the others. Married, and has two children, whose sight is perfect.

*(8) Female, æt. 25. (Mary F—, Case 1.) Nystagmus, colour-blind, and amblyopic. Tall, thin, very fair skin with freckles, hair light reddish-brown and abundant.

(9) Male, died at the age of two years of "whooping-cough."

*(10) Male, æt. 22. Sight very bad, worst of all, but the defect reported to be of the same kind. Very light brown hair.

*(11) Female, æt. 20. (Kate F—, Case 2.) Symptoms identical with those of No. 8 (=Case 1).

Parents have good sight and no colour defect, but an uncle is reported to be colour-blind, and unable to distinguish red from green.

Family II. Case 3.—This is an isolated case in a family of five children. Master S. G—, aged about 16 (Mr. Hutchinson's patient). Intelligent and fairly grown; teeth perfect. He is the only one of five whose sight is defective. His sight

has never been better than now, and formerly the nystagmus was much worse than it now is.

June, 1878.—V. each $\frac{12}{100}$; M. $\frac{1}{20} = \frac{12}{70}$ or $\frac{12}{50}$; no material astigmatism. Reads smallest print at 10″. Constant minute lateral nystagmus. Sees better in dull than in bright light, and always wears dark glasses in the daytime for this reason; even artificial light must be duller for him than for other people or he cannot see well. He is almost totally colour-blind, but says spontaneously that in very bright sunlight he can see red. In the spectrum he cannot distinguish any colours, only different shades. It is "lightest" at the red end.

Trial with Stilling's, Snellen's, and other tests showed that he could not distinguish with any certainty between any colours except by their luminosity; red he usually named correctly when bright or when held very close; yellow, blue, and green are indistinguishable except by differences of shade.

Ophthalmoscope.—Media clear; M. about $\frac{1}{20}$. Discs rather pale; the right showing a white, semi-transparent, glistening band of fibrous tissue, extending beyond its margin in the form of a capital D, and beneath the large vessels; the left slight haze along the large vessels. At the Y. S. region of the left a single small round spot of choroidal disease (a "punched out" spot), and in right a few similar, but much smaller dots, seen by the erect image. These appearances seemed to point to the previous occurrence of a choroido-retinitis, possibly just after birth.

The only important facts in the history were as follows:—The mother told me that three or four days before the patient was born the "waters broke and came away," and that the confinement was rather quick. When born the child was scarcely alive, and was indeed laid aside for a short time as dead, only being revived by artificial respiration. For several days he hardly opened his eyes, and seemed not to see; and then, when open, there was for several weeks "a sort of red skin" over the eyes, which, however, the medical man did not think important. The patient is the only one of Mrs. G—'s five children about whose birth and early infancy there was any peculiarity.

Family III.—Cases 4, 5, 6, and 7 are four children of one family (P—), with eye symptoms very similar to those in

Cases 1—3, but in addition with defective intellect, amounting in two of them to imbecility.

Case 4.—Philip P—, æt. 15, very tall and big for his age, but decidedly childish and backward; light hair, but irides almost brown. Rapid nearly vertical nystagmus of small range; pupils act well to light and atropine; no ophthalmoscopic changes whatever, but choroid highly pigmented. V. $\frac{20}{200}$, and with + 8 reads 8 J. at 4″; refraction H., but degree not estimated. Sees best in the evening and in the shade. Totally colour-blind; "I can only tell between white, black, and brown;" asked for "brown" he picked up scarlet. Confuses between bright yellow, blue, pink, and full dark green, and between full dark green and scarlet. The blues and yellows always look disproportionately light to him.

Case 5.—William P—, æt. 13, well grown; light brown hair; grey irides; rapid vertical and rotatory nystagmus. Pronounces "Th" as "F." V. in daylight $\frac{20}{200}$, and with + 8 = 6 or 8 J. badly at 2½″. Sees better in the dusk. No ophthalmoscopic changes. Total colour-blindness, his behaviour with the wools being identical with that of his elder brother.

Case 6.—Gertrude P—, æt. 7, is quite idiotic, and appears to be quite blind. I could not make an ophthalmoscopic examination.

Case 7.—Ernest P—, æt. 3½. Idiotic, and so nearly blind that he can apparently only tell between bright light and darkness. Constant nystagmus of wide range. Pupils usually 3½ mm., but slowly dilate to large size when he sits for some time in the shade. Discs dirty yellowish-white. Complexion very fair. Thickening of one side of septum nasi. Can walk and talk a little.

Family IV.—Cases 8, 9, and 10 are the three children of Mrs. C—, a first cousin of Mrs. P— on the mother's side. Her children are, therefore, second cousins of the P— children, whose cases have just been given.

Case 8.—F., æt. 6. Was seen at Moorfields by Mr. Streatfeild and others when a year old. There is now slight nystag-

mus, which is said to be diminishing; the mother does not know exactly when it began, but is certain the child took notice quite well till it was about three months old.

Refraction H., but degree not measured. V. = letters of 12 J. held close (daylight); sees much better in the dusk, but in the daytime her mother dare not let her go out because she cannot see enough to avoid large objects.

No ophthalmoscopic changes made out, discs of good colour, and no retinitis pigmentosa. Total colour-blindness; she matches pale green with pink, blue, yellow, and grey. Intellect precocious, her mother considering her "too forward."

Case 9.—Æt. about 4. Nystagmus much more marked than in Case 8, and V. defective. No ophthalmoscopic changes made out; discs perhaps too red.

Case 10.—Æt. about 12 months. Nystagmus of about the same degree as Case 9; can see. No ophthalmoscopic examination.

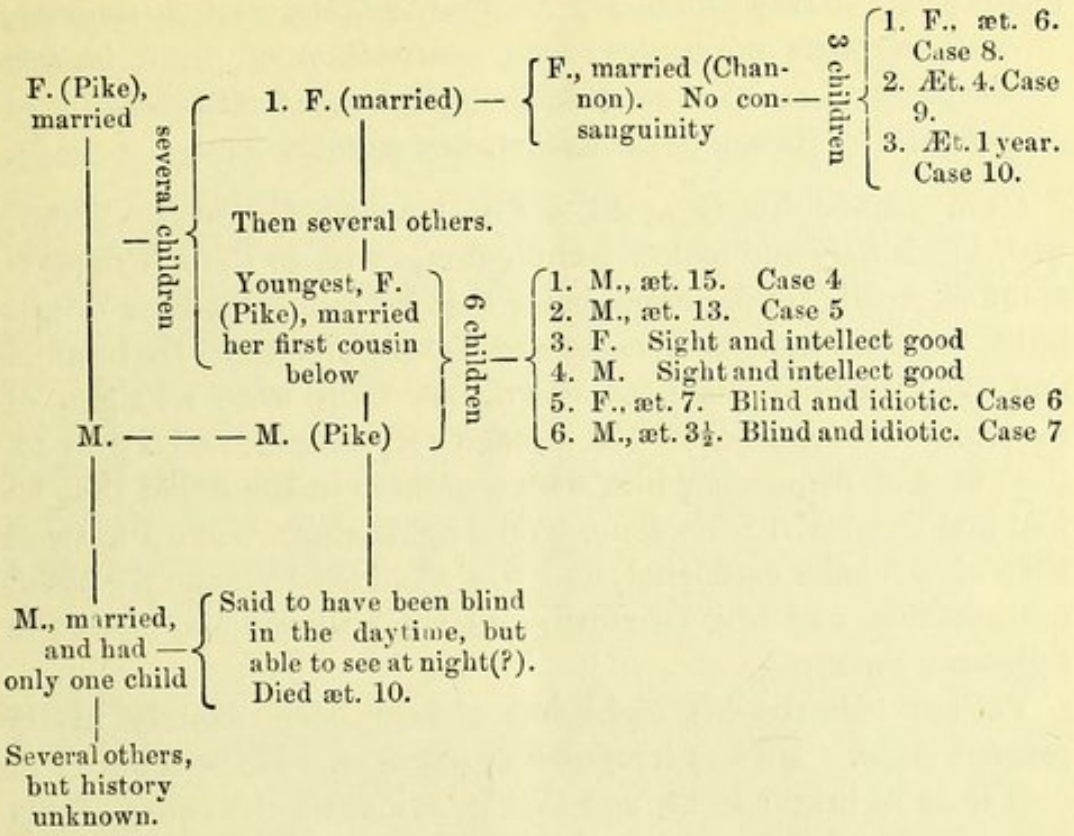
These two families of second cousins (III and IV) are especially interesting in connection with the influence of consanguinity of parents on the occurrence of defects in their children.

The children of the first family were the offspring of relations; Mrs. P— having married her first cousin; but the dependence of the disease on parental kinship is made much less probable by the occurrence of the second series in the family of Mrs. C—, where the parents were not related.

It is of course possible that there may have been consanguinity between the common ancestors of both families, and the unfortunate imperfections of the family history made it impossible to disprove this.

At a late visit Mrs. P— told me that a first cousin of her's, an only child which had died in childhood, was reported to have had bad sight in the daytime, and to have seen well at night.

The following table shows as much of the pedigree of these two branches of the family as I could collect:



Case 11.—Isabella Gleeson, 17, was in the Magdalen Ward at St. Thomas's in June, 1878. The history was imperfect, but in all probability her sight had been in its present state nearly all her life. V. less than $\frac{20}{200}$ with —20, which is the glass she chooses.

No ophthalmoscopic changes.

She stated spontaneously that her sight was best on dull days and in the dusk, and that she could not tell colours unless they were very bright. I did not test her completely, and can only say that she had defective perception of green, and probably to a less degree for red and violet.

She is one of a family of ten children, and two others besides herself are known to have bad sight.

Postscript.—Since the above cases went to the printer I have met with another family of which two members, if not three, suffer from *day-blindness and colour-blindness,* dating from very early in life, *combined with the ophthalmoscopic changes of true retinitis pigmentosa.* These cases are so extraordinary and of such interest in the present connection that I give the notes in detail.

FAMILY VI.—*Day-blindness, colour-blindness, and nystagmus, with retinitis pigmentosa and contraction of field, in two brothers; a sister said to be affected. Consanguinity of parents. History of melancholia in mother.*

CASE 12.—John G—, 23, a tall, coarse-featured milkman, with black hair and sallow complexion, came to the eye department in April, 1880, complaining that he could not bear bright light, and that his eyes became worse every summer. He blinked and liked to hold his head down, but there were no signs of external inflammation. He seemed very stupid, and I was on the point of dismissing him with a *placebo* in the belief that he had just recovered from some trivial ophthalmia, when I noticed that his eyeballs oscillated, and was thus led to make a careful examination, and Mr. Lawford, under my directions, made the following notes:—

Patient believes his sight has always been exactly in its present state. He has irregular nystagmus. He sees better in dull than in bright light, and this is evidently the cause of his fear of the bright light of summer; repeated and varied questioning fails to shake his statements on this point.

V. R. $\frac{20}{200}$, and 14 J. badly } Not improved by
V. L. $\frac{20}{100}$, and 14 J. badly } any glasses.

Visual field.—By the finger test, the fields are exactly symmetrical; they show great contraction in every direction except over a narrow tract in the downward and outward direction. Perimetrical examination of the right confirms this and defines the field as follows:

R.	Upwards	10°.
	Up.-in.	10°.
	Inwards.	10° or 15°.
	Down.-in.	15°.
	Downwards	15°.
60°	Down.-out.	20°.
30°	Down.-out.	90°. (very narrow tract.)
	Outwards	20°.
	Up.-out.	15°.

The boundary was sharply defined and constant with test-objects of various sizes, except in this down-out tract, where, in

order to gain uniform replies, I had to use a large test-object.

Colours.—Considerable deficiency of perception. When asked to name coloured wools he calls green "blue," and names red, yellow, and blue correctly. But he confuses purple with dark green, yellow with pink, and light blue with pink.

Ophthalmoscopic examination.—Pupils atropised. Retinal arteries decidedly small, and optic discs rather pale, but not at all "waxy." Abundant pigment, in very characteristic bone-corpuscle shapes, all round the periphery, except over a narrow tract at inner side, which, as far forward as can be seen, is perfectly free; this is quite symmetrical in each eye, and corresponds exactly with the tract of normal visual field. Media all normal.

Case 13. (*Notes by Mr. Davidson.*) Edward G—, æt. 25, milkman, elder brother of the above, attended at request for examination. Has much the same symptoms as his brother. Says he sees better in dull light. Is more intelligent than his brother, and very fond of reading. Has irregular nystagmus. Pupils act sluggishly.

V. R. $\frac{20}{200}$, and 6 J. at 8″ } Slightly improved by +30 or
V. L. $\frac{20}{200}$, and 6 J. at 8″ } 36, but not above $\frac{20}{70}$.

Colours.—Patient is aware he has colour-blindness. He confuses light green with bright yellow, pale greys, pale blue-green, pale buff, &c., and rose with dark blue, dark brown, orange, pink, and yellow.

Visual fields (abridged from detailed perimetric map) show very marked concentric contraction as follows in each eye.

Upwards	R. 15°	L. 18°.
Up.-in.	„ 18°	„ 18°.
Inwards	„ 20°	„ 18°.
Down.-in.	„ 18°	„ 18°.
Down.	„ 15°	„ 25°.
Down.-out.	„ 28°	„ 25°.
Outwards	„ 18°	„ 20°.
Up.-out.	„ 18°	„ 20°.

Ophthalmoscopic examination (*no atropine.*)—Some pigment

at the periphery, especially towards lower part of retina; the amount is about the same in the two eyes.

Case 14.—Mary G—, æt. 34 (not seen), is said to be affected in the same way as her two brothers. She cannot see so well in bright light, and cannot face the light. "Her eyes move about very much." She is believed to see colours wel. She is married and has two children living, whose sight is believed to be very good.

From the mother, who came at request, I obtained the following facts:—She and her husband were first cousins once removed. The husband died of heart disease aged 50. She herself is a nervous melancholic woman; married when 22 years old, and about the time the two amblyopic sons were born seems to have suffered from melancholia for a considerable time. She has had *nine* children, of which the *first, sixth,* and *seventh,* have imperfect sight. The remainder, of whom all are alive but one, are believed to have good sight. The defect of sight was noticed in all three when very young.

The pedigree is as follows:

M. (Gould), married.

M. — M. — F. (Gould), melancholic

married 1st cousin once removed

M. — M. (Gould), died of heart disease.

9 children.

*1. F., 34. Said to be affected (Case 14).
2. M.
3. F.
4. F.
5. F. Dead.
*6. M., 25 (Case 13).
*7. M., 23 (Case 12).
8. M.
9. F.

I have decided to offer pleasure resembling Cases 1 to 11.

(1) 2 Sisters (Germans) in a family of 4; Their parents were 1st Cousins.

(2) 2 Sisters (sent to me by my friend Mr. Marcus Jay) in a family of ; Their parents were also 1st Cousins