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HEREDITY IN RELATION TO THE EYE.*

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A great and steadily increasing interest in the ocular effects of heredity has arisen in recent years, and much knowledge has been acquired by the collective work of many investigators of this hitherto neglected subject. Of all the patient and productive workers in this field Edward Nettleship is far in the lead. His investigations have included the following hereditary diseases: Cataract, retinitis pigmentosa, congenital stationary night blindness, hereditary optic atrophy, color blindness, glaucoma, iritis, chorioiditis, opacity of the cornea and nystagmus. Not only has he investigated personally many affected lines from two or three even to ten generations, but he has also tabulated the work of ophthalmologists the world over; thus giving the comprehensive observations and statistics upon which alone intelligent opinion may be founded as to this important factor in ophthalmic science.

The revival of interest in Mendel's rules on heredity and the application of them to the transmission of anomalies, deformities and diseases in man, have doubtless stimulated ophthalmologists to investigate along these lines in their own particular domain.

Congenital defects not traceable to the same or similar abnormality in the direct line, have not been included in the following summary of illustrative cases.

LIDS.

The writer has observed hereditary narrowing of the palpebral aperture of one eye as a family trait, extending for three or four generations. Other familial peculiarities may be noticed by careful observation of generations known in person or by study of family portraits or photographs.

Epicanthus. Double epicanthus running through two and three generations is fairly common. In each instance the development of the bridge of the nose corrected the deformity by or before adult life so that operation seemed inadvisable, in cases observed by the author.

Coloboma of the lid has been observed as an hereditary anomaly.

Ptosis. In a personal communication, Samuel Z. Shope has related a striking case of hereditary ptosis, affecting the right eye in


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five generations. One child only in a large childship was affected in each generation. A female showed this defect in the fourth generation of the affected line; males only being affected in the other generations.

H. R. Stilwill has mentioned to the writer a case of ptosis affecting a man, his daughter and her son.

Ectropion. Harry A. Smith has personally told the author of ectropion of both lower lids, occurring after forty years of age, which he had observed in a father and his seven sons.

Cornea.

Family Degeneration of the Cornea. Buchanan (1) reported the case of a woman, aged thirty-six, showing this defect, and completed the previously noted histories of her two brothers, who had the same disease. One brother died of tuberculosis. The vision of the other gradually failed until he had to give up his occupation as an engine fireman. In the sister's eyes both corneas were similarly affected. The central area was distinctly hazy for a radius of about 2 m. m.; around this was a ring of dotted opacity 1.5 m. m., and beyond this a clear ring to the periphery. The corneal microscope showed the central haze to be made up of very small bluish-grey dots. The vision was 4/36 in each eye, the tension and pupils normal, and the health said to be good.

Folker (2) observed family degeneration of the cornea in three generations, eight cases ranging from twelve to ninety-two years of age. All the affected areas were nodular, and both eyes were involved in each case.

Komoto (3) noted a father, son, daughter and nephew, who were all born with diffuse, non-vascular corneal opacities of a bilateral character. The son and daughter had congenital cataract, also. Neither syphilis nor tuberculosis could be detected.

Lattice-Form Opacity. Freund (4) reported fifteen cases of lattice-form opacity of the cornea, occurring in two families, and running through at least four generations. The opacity appeared at or after puberty, reaching its full development between thirty and forty years. Both eyes were involved, but not to the same extent. The opacity was thickest at the center, with a clear periphery. Both the superficial and deep layers were affected; and a chalky deposit appeared in late stages. No constitutional disease was detected.

Interstitial Keratitis. This disease is usually considered as pathognomonic of inherited syphilis, although a small number of cases are due to acquired syphilis, and probably fewer still to inherited gout.
The writer had under his care a case of interstitial keratitis due to acquired syphilis, and following the primary infection by only nine months; and has reported cases due to hereditary taint. In one of these inherited cases the mother of the affected child took active and persistent anti-syphilitic treatment for three years preceding marriage, the last six months of pregnancy and the first three months of nursing her first child. When six and one-half weeks old this child had a skin eruption on each heel, which yielded to mercurial inunctions in two and one-half weeks. Snuffles were persistent for the first six months of the child's life. At four years of age double interstitial keratitis of a severe and distressing type developed. The corneal inflammation subsided in two months under the use of atropin and heat locally, and calomel and hydriodic acid internally. In two months more the opacities in the cornea disappeared under applications of yellow oxide of mercury ointment, alternated with dionin.

Parenchymatous keratitis is sometimes much modified in severity in the second eye affected; due, apparently, to active specific treatment during the first attack.

Sclera.

Blue Sclerotics. Adair-Dighton (5) has reported blue sclerotics in nine persons, running through four generations, and transmitted through males only. Six out of eight children in the third generation had this condition; and the only member of the fourth generation yet born also showed it. Osteoporosis, as shown by frequent fractures, was generally manifested in these cases. It was thought that the blue color was due to thinning of the sclerotics.

Sydney Stephenson observed blue sclerotics affecting twenty-one of thirty-two members in four generations of a family of syphilitics. Harmon followed this genealogy further, finding thirty-one out of thirty-five showed some congenital peculiarity. Rolleston (6) reported a syphilitic mother who, with her male infant of five months, had blue sclerotics. Her sister and grandmother were likewise affected.

Iris.

Aniridia. Report has been made by Hamilton (7) of a father and three sons who showed congenital absence of the iris. The mother was highly hyperopic, but her eyes were otherwise normal.

Hereditary Defect of Iris. Thye (?a) reported bilateral congenital defect of the anterior layer of the iris affecting father (aged 32 years) and son (aged 10 years) similarly, although the lesion was more extensive in the father's eyes. The other living
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child, a girl, had normal eyes; but an infant that died at six months had the same anomaly as the father, according to the statement of the observing and credible parents. Both father and son were physically sound except as to their eyes. Both had horizontal nystagmus, and the son had alternating divergent squint also. The father showed operative aphakia in one eye, mature cataract in the other. The son presented punctate opacities in the left lens. The anterior layer was absent from about one half the surface of the father’s irides, and about one-fourth of the son’s. The pigment layer showed distinctly in this area; and in one spot in one eye of father and son the iris was entirely deficient, the lens opacity being seen through these openings. Both patients were decided blondes; and the gray-blue of the unaffected stroma made a sharp contrast to the brown, exposed pigment layer of the iris.

They also referred to Manz’s case of hereditary bridge-coloboma affecting two generations.

Lenses.

Cataract. Nettleship (8) has traced senile and juvenile cataract through one hundred sixty-seven families, three to six generations being affected; and has studied two hundred thirty-eight cases of congenital cataract. In one family thirty members in four generations showed the defect. The transmission was direct, from one or both parents to offspring, rarely skipping a generation. In some families he noted a striking tendency for the cataract to appear at about the same age, generation after generation; but on the whole it appeared or ripened earlier in the later generations. In four out of ten families affected; the parents of children with complete congenital cataract were first cousins.

Nettleship and Ogilvie (9) have together collected twenty cases of congenital cataract among 150 members, in a family history covering seven generations. The time of appearance of the cataract ranged from 10 to 82 years. In every case the inheritance was direct, from parent to child.

Millikin (10) observed fourteen cases of hereditary cataract in three families. In two it was traced through three generations. In one family cataract developed during early childhood for three generations, being bilateral in each case.

Collomb (11) has reported zonular cataract in two, and probably three generations. In six children of the supposedly third generation one was slightly affected, while two showed pronounced cataract, and three were unaffected.

Manson (12) has recorded a case of hereditary lamellar cataract
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occurring in four consecutive generations, affecting six males and seven females, and transmitted in all cases by affected females. Three members of the pedigree in different generations had a congenital deformity of the little fingers; but only one person showed both the digital and lenticular abnormality.

Posterior Polar Cataract. W. F. Matson has recently observed binocular posterior polar cataract affecting two daughters of a man who had experienced defective vision all his life, but without a history of traumatism or inflammatory disturbance. One of these daughters had no children; the other had two daughters and a son, both girls showing the mother's defect in each eye.

Hereditary Punctate Opacities. The writer has observed numerous peripheral punctate opacities in the outer fourth of the lens, of identical appearance, in a mother, aged thirty-seven, and her daughter of twenty years. Except for errors of refraction these eyes were not otherwise affected. Correcting lenses gave normal vision.

Ectopia Lentis. A. R. Gunn (13) reported seventeen out of twenty-two children, the offspring of an affected and unaffected parent in five families, all of whom showed bilateral and complete congenital dislocation of the lens, without coloboma of the uveal tract. The lens was well developed and clear in the three children examined, and floated in the vitreous. In three adults examined, small pupils and posterior synechia prevented a satisfactory inspection of the interior of the eye, but the absence of the lens was demonstrated.

G. G. Lewis (14) noted hereditary dislocation of the lens affecting sixteen persons in six successive generations.

Congenital Aphakia. Toufesco (15) has reported a case of congenital aphakia, and collected fifteen other cases previously recorded. Most of the cases showed other abnormalities of the eye. Toufesco expressed the belief that, as a rule, the absence of the lens was due to a process of degeneration and absorption of the previously formed lens, rather than a fault of embryonic development.

Optic Nerve.

Hereditary Optic Atrophy was first described by Leber (16) in 1871, and is called "Leber's Disease." It is an affection of the papillo-macular bundle of neurons, is evidenced by a central scotoma and ends in optic atrophy of varying degrees. Before Helmholtz invented the ophthalmoscope, in 1851, these cases were doubtless recorded as hereditary amblyopia or hereditary amaurosis.
In 1877 Leber added to his previously reported cases, collecting a total of fifty-five cases in sixteen families.

Behr (17) described an unusually early appearing bilateral optic atrophy which he classed as hereditary. It occurred in six cases, and all were boys, affected in early childhood. Other parts of the nervous system were involved.

Raymond and Koenig (18) have recorded four family groups of optic atrophy, and point out their medico-legal importance. They cite cases in which compensation was claimed and allowed for optic atrophy ascribed to slight injury or to retrobulbar neuritis due to exposure.

Hanke (19) had made a similar observation in the case of two brothers.

Batten (20) reported two cases in one family, together with a cousin. All were affected before twenty. The family history indicated transmission through healthy mothers, as is usual in this disease.

Twelve cases of optic atrophy in one family were traced by Hancock (21) through five generations. Six cases recovered good sight. Each affected person was a male, and the optic degeneration was transmitted through healthy daughters to grandsons of the affected male.

In 150 cases atrophy appeared between 6 and 67 years, the largest number occurring at about 20 years.

Arnold Knapp (22) observed eight cases in three generations, being discovered at six years of age. The disease was transmitted from the father to two sons and two daughters, and through one of these daughters to three of her sons.

In 1909 Nettleship (23) collected all published and many unpublished cases of this disease; in all 180 separate records with references.

**Retina.**

**Glioma.** De Gouvea (24) recorded an instance of heredity in glioma. The father had the right eye removed when two years old, for glioma. This diagnosis was confirmed by the microscope. At twenty-one he married a woman whose family history was good in reference to neoplasms. They had seven children. Of the first two, both girls, one had glioma of the retina at five months, the other at two years. Another female child developed glioma at five months. The other four children escaped this form of malignant sarcoma.

**Retinitis Pigmentosa.** Of 488 families affected by this disease,
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which is incurable and usually leads to practical blindness, Nettleship (25) found heredity without consanguinity in 230, consanguinity without heredity in 226, and both these factors figured in 32 affected families. It has not been proved that consanguinity can originate retinitis pigmentosa, as Liebrich thought, even though the offspring of cousins show it with striking frequency; but intermarriage does surely act as an intensifying cause by increasing an hereditary tendency. A family has been reported in which twenty males and eighteen females were affected by retinitis pigmentosa in seven generations. The disease has skipped even three generations and then reappeared with undiminished force.

Snell (26) has traced a family history of 67 descendants, 28 showing night blindness and other evidence of pigmentary degeneration of the retina; males and females being affected in about equal proportions. The disease affected five generations, skipping none. It was transmitted through both males and females, and consanguinity was not proven. Night blindness made its first manifestations in early childhood, and was complete at 40 years.

Aubineau (27) has reported three out of five of the children of second cousins affected by retinitis pigmentosa; and the case of two affected brothers, whose parents were first cousins. The averages of many observers who have collected data indicate that consanguinity is a factor in fully 25 per cent of all cases of retinitis pigmentosa.

Posey had under observation two generations of a family affected by retinitis pigmentosa, it is said, for two centuries. Posey and Sautter (28) have urged that consanguineous marriages should be discouraged by the profession and prohibited by law.

Nettleship has called attention to one type of heredity which affects the sexes in about the same proportion, and another in which the males only are affected but the disease is transmitted through the female line alone. He considers that consanguinity of parents emphasizes and even originates hereditary taint. It has also been noted by him that certain families seem subject to varying diseases of the eye, one member having glaucoma, another cataract, a third detachment of the retina or gouty inflammation; and that probably this sometimes indicated an hereditary imperfection of the whole eye, comparable with certain defects of the nervous system.

General Hereditary Diseases.

Glaucoma. Nettleship (29) has suggested the probable disproportion in size between cornea, ciliary region and lens in inherited pri-
mary glaucoma. Three to five generations have been affected; the most extensive invasion being nine out of twenty-two persons in two generations. Anticipation, i.e., the tendency for the disease to appear at an earlier age in succeeding generations, is very marked in some of the cases of glaucoma where heredity can be proved. The child may have both compensatory myopia and glaucoma.

Ametropia and Squint. Myopia, hyperopia and astigmatism are apparently hereditary defects. Myopia in one parent may be overcome by hyperopia in the other. A case comes to the writer's mind of eight diopters of myopia in the mother being counteracted, as an hereditary influence, by emmetropia or hyperopia in the father's line, so that the four children of these parents were emmetropic. Fleischer (30) found, in a German village where myopia was especially common, that fifty per cent of the children in seventeen families in which one parent was highly myopic, developed myopia. In another family in which both parents were myopic, all the children manifested the same error. The parents of eighty families in this village had normal eyes in each instance, as did their offspring.

Different authorities have variously estimated squint to be hereditary in from 33 to 70 per cent of all cases. Heredity is especially marked in convergent strabismus.

Von Sicherer (31) traced squint through four generations of one family.

Based on ophthalmometric measurements in a large number of children and one or more near relatives, Steiger (32) found corneal astigmatism inherited from one or the other parent in a large proportion of cases.

Czellanitzer (33) analyzed 330 families affected with more than 6 dioptres of myopia. Male and female members appeared to be equally involved as to the hereditary factor. In 30 per cent of the cases there was myopia of both parents, in 20 per cent of the father alone, and in 17 per cent of the mother alone. Transmissibility of myopia seemed to diminish in later children.

Bogatch (34) has reported three generations of a family of thirty-eight members. Eleven had high myopia; some with retinal detachment. In all the high myopia developed in early youth, with early blindness in some cases. Consanguinity was apparently a factor, as the grandparents and parents of the most affected children were cousins. There was also an hereditary predisposition to myopia from diminished resistance of the sclera at the posterior pole of the eye.
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Ophthalmoplegia. Bradburne (35) has traced this rare affection through five generations. He found ptosis, accompanied by an almost complete loss of ocular movements.

Huetlemann (36) published his observations on congenital ptosis, with epicanthus, in three generations. Eight children out of eleven were affected. In only one case was there disturbance of other ocular muscles. The electric current showed absence or imperfect development of the levator.

Nystagmus. Clarke (37) has noted this disease extending through five generations, affecting 23 persons, all males. In each generation none of the daughters were affected; and only the eldest daughter transmitted the defect, and she to her sons only.

Radloff (38) described a new family group of miners' nystagmus, which is characterized by very regular undulatory oscillations, varying from 180 to 240 per minute. Vestibular nystagmus, on the contrary, is of the jerking type.

From thirteen pedigrees of hereditary nystagmus Nettleship (39) discovered that in those showing both eye and head movements the abnormality affected both sexes, and could be transmitted by either male or female parent; whereas, in the genealogies showing eye movements alone, the abnormality was strictly limited to males and transmitted only by unaffected mothers.

Amaurotic Family Idiocy. R. M. Smith (40) has reported two cases occurring, as usual, in Hebrews. The characteristic symptoms are a mahogany red spot on a white background in the macular region, decreasing vision, progressive paralysis, mental deficiency going on to idiocy, nystagmus and drooling. All die before three years of age.

Sheffield (41) has recorded the case of a Hebrew child of eleven months. It developed well the first six months; when it gradually became pale, flabby, less active physically and mentally, and developed the characteristic fundus changes, with blindness and idiocy. The child died of grippe and pneumonia. An older brother and sister were normal.

Hereditary Blindness. Clarence Loeb (42) has reported a family in which every member for five generations was affected with cataract, and also six families, headed by blind parents, in which seventeen out of thirty-one children were blind. He thinks ten per cent of blindness may be hereditary. In optic atrophy he noted a marked tendency to transmission through a healthy mother to a blind child. It is his opinion, and that of a large number of ophthalmologists to whom he addressed inquiries on this subject,
that the marriage of a person afflicted with hereditary blindness should be advised against and, if possible, legally prevented.

The United States census of 1900 showed that 56,507 inhabitants were blind. Of this number 2,527 were the children of cousins. Bemis traced 823 marriages of cousins, finding 85 blind (over 10 per cent), and 145 deaf mutes.

*Color Blindness* has been frequently traced as an hereditary influence.

**Congenital Stationary Night Blindness.** This striking affection has been traced by Nettleship (43) for 270 years in the Nougaret family, of Southern France. Out of a genealogy of 2,121 members, 6.36 per cent were affected. The disease shows no pigmented or other retinal changes. The general health is good and longevity is noticeable. Invariably this condition is transmitted directly from night blind parent or parents to offspring. The tendency to night blindness is emphasized by frequent intermarriage.

Sinclair has collected eleven cases in a family of forty members, the disease affecting four successive generations.

Nettleship (44) has also reported twenty cases of congenital night blindness in which this condition was associated with myopia, and descended through normal females.

Langdon (45) has reported a family of five in which the father and one daughter were affected by more or less helplessness in dim light. Corrected vision was 6/5, the fundi were of healthy appearance, and the fields were normal for form and color in a good light, with concentric contraction in diminished illumination. Henry's photometer showed the daughter's light sense to be R. 3/5 and L. 2/5, while the father's was 3/5 in each eye.

Bordley (46) has noted congenital night blindness in a group of cases traced to five generations. It was associated with defect of the lower temporal quadrant of each visual field. There was progressive concentric contraction of the fields, which were greatly narrowed by diminished illumination. Death occurred in middle life, about one year after total blindness became established.

**Congenital Word Blindness.** This defect, which is cerebral rather than ocular, has been traced by Stephenson through three generations. Hinshelwood reported four cases and pointed out in 1907, that children with this defect never get a fair chance in public schools, but do very well with private instruction, when not subject to ridicule for their great backwardness in learning to read. In all respects but retention of the visual impression of words, the mentality is normal. Four years later Hinshelwood
reported two cases occurring in the second generation of that same family. One of these cases showed defective visual memory for both words and figures; though this investigator has shown, from his study of cases of acquired word blindness that the visual memory for words and letters is completely independent of that for figures. All of Hinshelwood’s many cases of congenital word blindness have ultimately been taught to read; showing that none were defective as to general intelligence.

Albinism. Lagleyze (48) found 27 cases of albinism, mostly among people in an isolated province in the Argentine Republic, where intermarriage had been practiced to an extraordinary extent for nearly a century. These 27 cases occurred in 13 families; and 5 of these families produced 13 cases in children of first cousins, or of uncle and niece. Lagleyze considers that consanguineous marriages are even more of a factor than heredity in causing albinism, and that a parent with partial albinism may have offspring showing complete albinism. In this way hereditary deficiency of pigmentation is intensified.

Cryptophthalmos. Coover (49) has reported a mother and child with bilateral congenital cryptophthalmos. Since that report another child has been born with the same anomaly. The father and four grandparents of these two children showed no such defects.

Inheritance of Acquired Ocular Defects. This question is of interest, even though it has not passed the speculative stage. Tobias (50) has recorded the instance of a mother with bilateral operative colobomata of the iris who gave birth to five children, the two youngest showing congenital coloboma of iris and choroid. The eyes of the other children were normal. The operation on the mother had occurred four years before her marriage. In her right eye the coloboma was below and in, in the left, up and in. One child had bilateral coloboma below, the other showed a coloboma of iris and choroid down and in. Failure of the fetal cleft to close may have caused the congenital colobomata, rather than direct inheritance of the results of surgical interference.

More cases could be related along the lines considered, and even different manifestations outlined. But the part that heredity plays in the transmission of ocular disease seems already proved and sufficiently illustrated. It becomes, therefore, the duty of physicians in general and ophthalmologists in particular, to use their utmost endeavor to discourage the marriage of a person or per-
sons afflicted with hereditary ocular disease, and if possible to secure the legal prohibition of such alliances.

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