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PART I.

ORIGINAL COMMUNICATIONS.

ART. VII.—*On Coloboma of the Chorioid and of the Optic Nerve Sheath.*^a By ARTHUR H. BENSON, M.B., F.R.C.S.I.; Assistant Surgeon, St. Mark's Ophthalmic Hospital; Lecturer on Ophthalmic and Aural Surgery in the Ledwich School of Medicine.

THE cases of maldevelopment of the internal tunics of the eye which I have the honour of exhibiting to the Medical Society are of interest, not alone from their extreme rarity, but also from an embryological and practical point of view.

All maldevelopments or arrests of development help to teach us the mode of development, or at least serve as useful means of correcting theories and conclusions otherwise deduced. To the practical physician also it is of importance to remember that such malformations do occur, in order that he may not mistake them for the effects of disease, and perhaps suggest treatment which must fail to benefit.

Both cases were discovered whilst assisting my friend, Dr. Fox, of Philadelphia, to examine, for statistical purposes, the eyes of the deaf-mutes in the institutions about Dublin. Both are deaf and dumb—the boy being an inmate of St. Joseph's Male Asylum at Cabra, and the girl of St. Mary's Asylum, also near Cabra. Neither was known to have anything amiss with the eyes until

^a Read before the Medical Society of the King and Queen's College of Physicians, Wednesday, February 1, 1882. [For the discussion on this paper see page 226.]

we discovered the colobomata. The following are the notes of the cases:—

CASE I.—J. H., aged thirteen, otherwise fairly well developed and intelligent, was admitted into St. Mark's Hospital in October last. There is nothing remarkable about the external appearance of the eyes; in both the iris is normal in shape, and acts well to light and accommodation. The tension also is normal, and the lens is clear. On examining with the ophthalmoscope the media are found to be perfectly transparent, the disc is normal in shape and colour, with considerable pigmentation round its border, and the general fundus is also normal, though slightly mottled. The refraction is hypermetropic to the extent of two diopters. R., V.= $\frac{6}{12}$??; L., V.= $\frac{9}{12}$??.

However, on investigating the condition of the inferior segment, a large, almost circular, white patch with pigmented borders is seen occupying a position below and somewhat to the inner side of the disc in each eye, and about midway between it and the ciliary body. It is larger in the right eye than in the left, but the positions are symmetrical. Retinal blood-vessels are seen running over the surface of the patches, the edges of which are elevated and darkly pigmented in places. As these vessels, which can be traced directly to the central vessels of the disc, cross the border of the coloboma, they give a sudden bend as over the edge of a steep glaucomatous cup. The floor of the coloboma is myopic to the extent of two diopters, making the difference between the level of the general fundus and the colobomatous area equal to four diopters, or one millimetre in antero-posterior measurement.

In the right eye the inferior border of the coloboma is somewhat pointed and not evenly pigmented; the floor of the coloboma is pearly white, with a few faintly pigmented patches scattered over it. The chorio-capillaris seems to encroach a certain distance over the pigmentless area of the coloboma. In the left eye the inferior border is evenly curved, but at its most inferior portion a good-sized vessel bends round its margin, and runs towards the periphery of the fundus. This vessel cannot be traced over any portion of the coloboma, but on the retina its margins for some distance are marked by a line of black pigment. I regard it as an errant ciliary vessel, such as is not infrequently seen even in normal eyes, running to supply some portion of the retina.

The field of vision tested with Förster's perimeter is of almost normal extent in both eyes, but the acuity of vision is considerably impaired in the upper portion of each. No definite scotoma could be made out, but the boy said that the test object (a piece of white paper one centimetre square) was not so distinctly seen above as it was below or at the sides.

The difficulty of taking an accurate perimeter chart in a deaf and dumb boy is considerable, and would have been insuperable

but for the kindness of the Rev. Mr. Ennis, of the Institution in Cabra, who came to St. Mark's Hospital and made the boy very fully understand what was required of him, and interpreted his signs to me.

CASE II.—B. M. is a rather ill-developed and not very intelligent child, aged thirteen, deaf and dumb from her birth. All her brothers and sisters speak and hear normally. External examination of her eyes fails to discover any anomaly of the iris or other portions, and the media are clear; tension is normal, and vision in the right eye is about $\frac{6}{12}$, and in the left the same. The ophthalmoscope reveals a high degree of hypermetropia—viz., four diopters in the fundus of each eye. The chorioid and retina are normal, or perhaps unduly mottled in the periphery; but the appearance of the disc-region is truly remarkable.

In the right eye no disc, properly so called, can be made out, but in its place a large white, irregularly depressed funnel can be seen, from the lip of which the retinal vessels seem to take origin, and over the edge of which they bend sharply, as in a glaucomatous cup. This appearance is best marked below, the superior lip not being so abruptly terminated, but gradually receding on an inclined plane from the level of the healthy fundus into the deep excavation below. Tracing the large retinal vessels we find that above they pass over the pigmented edge of the coloboma without undergoing any sudden change in direction. Following them on, they gradually get more and more indistinct until they are finally lost in the depths below. Those to the lower outer side pass suddenly from view, disappearing as over the edge of a precipice, whilst those from the lower inner side, passing over the pigmented border, without change in appearance, run for a short distance and then suddenly disappear, again to become visible, but dimly, on a lower platform, where by their direction and size they can be recognised as continuations of the above. These again bending sharply over the edge of the ledge on which they run, also disappear into the hidden depths. The area of this pigmentless disc-region is about six times the size of the normal papilla.

Just below and to the inside of this disc-region, and separated from it by a narrow isthmus of fairly healthy fundus, is seen an unpigmented oval patch about one-fifth its size. This patch closely resembles in its appearance the patches seen in the eyes of the boy first mentioned. Over its surface, which is somewhat depressed below the level of the surrounding retina, several good-sized retinal blood-vessels run; its borders are slightly pigmented in places, but its surface is entirely devoid of pigment, and presents the pearly white appearance of the sclerotic. In its centre this patch shows a depression corresponding to a difference in refraction of three diopters as compared

with the surrounding retina. Calculating, then, that four diopters of refraction correspond to one millimetre in antero-posterior measurement, we find that this inferior coloboma is depressed three-quarters of a millimetre, and the upper coloboma of the optic disc has a depression of about two millimetres in its centre, judging by the position of the fine vessels which ramify over the pearly or opalescent membrane which seems irregularly to cover its floor, leaving still deeper holes behind. This membrane is probably retinal in origin.

The left eye, like the right, is strongly hypermetropic (4 D.), and the fundus is everywhere mottled. In it, too, the disc is entirely abnormal, being about twice its ordinary size, and instead of being circular, or evenly oval, it is peaked at its inferior extremity, making it more or less pear-shaped. As in the right eye, its surface is unevenly depressed; the vessels from above gradually disappear, whilst those from the inferior segment suddenly disappear over the edge of the precipice to be no more visible.

The depth of the depression here, estimated in the same manner as in the right eye, is about one millimetre.

There is no inferior coloboma in this eye, but in its place, and running downwards and inwards, is an irregularly pigmented band, with an ill-marked median line, suggesting the idea of a raphé or junction-line.

The field of vision in this case I also attempted to test with Förster's perimeter, but without success, as the child's education is not sufficiently advanced to enable her to understand the signs, &c., of such complex ideas as those necessary to explain what was required of her.

The accompanying drawings serve to illustrate the peculiarities in each case.

Through the kindness of my colleague, Mr. Story, I am enabled to show the drawing made by Dr. Knaggs, of Sydney, of a case (M. D.) of double coloboma of the chorioid and iris, which was under his care in St. Mark's Hospital last year. The right lens was cataractous, so that the condition of the chorioid and retina in that eye could only be conjectured; but in the left eye there was a very large pigmentless area, embracing the optic disc above, and reaching to the ciliary body below, where it joined with the split in the iris. The disc margins were very badly defined; the fundus was dark, and therefore contrasted the more vividly with the glistening white of the colobomatous area, over which retinal vessels freely ramified. The field of vision was everywhere restricted, but especially above, where it reached only to 23°. T. n., V. = fingers at 4 metres.

"Coloboma of the chorioid" is the name by which the affection

is most generally known, and which, therefore, I have retained, but only under protest, as the term is so loosely used to express widely different conditions.

Before going further, I may, perhaps, be permitted very briefly to sketch in outline the mode of development of the essential structures of the eye, so that the description of the cases may be the more readily followed.

Very early in the development of the embryo a protrusion forwards occurs on either side from the anterior primary encephalic vesicle. This prolongation or expansion, the so-called "primary optic vesicle," forms a hollow pedunculated chamber, communicating by its stalk with the general ventricular cavity of the primitive brain. As soon as the lens begins to develop by an involution of the cuticular epiblast, the anterior inferior portion of the primary optic vesicle becomes doubled back on itself, so as to form a cup-like depression, the "secondary optic vesicle" or "optic cup."

The superficial fold, or that towards the brain, rapidly diminishes in thickness, and produces only a layer of pigment cells, the so-called chorioidal, but, as we see, more properly named retinal pigment; the deeper fold, at the same time, increases in thickness, and forms the various elements of the future retina, whilst the pedicle becomes the optic nerve. The space which exists between the doubled back junction border of the two folds is the "ocular foetal fissure." Through this the mesoblastic elements gain entrance into the interior of the globe to form the blood-vessels and vitreous humour. This fissure extends back into the pedicle for a certain distance, thereby permitting the entrance of the central artery of the retina into the interior of the optic nerve. The closure of the foetal fissure normally takes place about the seventh week of intra-uterine life, whilst yet the mesoblastic coverings, the chorioid and sclerotic, are still in a soft and very rudimentary condition. It is with this fissure and its closure, or mal-closure or non-closure, that we mainly have to do in explaining the mode of origin of such malformations as those before us. The iris, which, according to Manz (Graefe and Sæmisch's Handbook), does not begin to develop until about the same time as the split in the secondary optic vesicle completes its closure, is partly derived from the anterior lip of the optic cup, and partly from the chorioid proper. It is normally developed without a fissure, as is the chorioid itself. If this be true, it is manifestly incorrect to speak of coloboma of the iris or chorioid as an arrest of development,

since such conditions exist at no period of normal embryonic life; they should rather be spoken of as malformations or maldevelopments. But opinions differ on this point, some observers having stated that they could see the split, even with the naked eye, in the embryo chick of five days.

For very many years a malformation of the iris in the form of a V-shaped gap or split—the apex of the split being at the ciliary border, the base at the pupillary edge—has been noted as a congenital condition, and described as “coloboma of the iris.” But it was not till long afterwards that Von Ammon, for the first time, observed a deficiency in the posterior tunics of the eye in conjunction with the iris split. As Burckhardt Wilhelm Seiler, in his work on “Malformations of the Eye,” page 44, published at Dresden in 1833, says:—“The split of the chorioid (coloboma chorioideæ) and of the retina, which, during the development of the eye, normally exists in the embryo, has been found by Von Ammon (*Zeitsch. f. d. Ophthalmologie*, I. B. S. 58) in an adult as an *arrest of development* (*Bildungshemmung*) in conjunction with coloboma of the iris.”

It would be tedious, and not very profitable, to go over *seriatim* the various opinions held by eminent authorities as to the nature and cause of the anomalous condition of the fundus oculi discovered by Von Ammon. The theory of its genesis held by its discoverer, and, until lately, by almost all observers, seems to have been based upon notions of the history of the development of the human eye, the correctness of which has since been questioned. To them the anomaly was simply one of arrested development analogous to hare-lip or cleft-palate.

Anatomical and microscopical examinations of colobomatous eyes have, however, demonstrated, in many cases, the presence of retina, more or less perfect, over the colobomatous areas; and Dr. Hermann Pause, in Graefe's “Archives of Ophthalmology,” 1878 (Vol. XXIV., Part 2), relates a case in which he examined microscopically the colobomatous eye of a new-born child, and found in the region of the coloboma “the retina, with all its elements, continued in the same order as in the normal membrane—neither a thinning nor a stretching of the individual components, nor even the presence of heterogeneous elements. Further, there was no abnormal adhesion with the chorioid, which was itself quite normal throughout; in particular, neither the elastic lamella nor the chorio-capillaris were

wanting." Even the pigment epithelium was present, but devoid of pigmented matter, so that the only abnormal condition found was a "leucosis of the pigment epithelium" of the retina over the affected area. Other investigators have found complete absence of retina and chorioid in the region of the coloboma, whilst Manz, in his article on Malformations of the Human Eye, in Graefe and Sæmisch's Handbook, states:—"The rule is that within the coloboma the retina is completely wanting" and that "besides this constant defect, the elastic lamella and the choriocapillaris are usually absent—structures which belong to the chorioid proper, and have no connexion with the foetal fissure." A simple arrest in the closing process of the secondary optic vesicle is, therefore, insufficient to account for these various anatomical conditions, and we must seek for other factors in their production.

To quote Manz again, there are three factors which in coloboma of the bulb are of importance, although not to the same extent in all cases—(1) a disturbance in the closing process of the foetal eyesplit; (2) a secondary disturbance in the development of the tissues derived from the cephalic plates which envelop the eye (viz., chorioid and sclerotic); and (3) an ectasia of the resulting cicatrix and of the neighbouring structures, the result of intra-ocular pressure. The contraction of the cicatricial tissue with which the foetal fissure gets closed may have considerable effect in diminishing the size of the area, at first devoid of retina, and may account for many of the anomalous microscopical conditions described by authors.

As to the nature of the disturbance which primarily affects the closure of the foetal fissure we know nothing more than that it must come into operation before the seventh week of embryonic life, for that is the time at which the secondary optic vesicle closes. There is, however, some evidence in favour of its being inflammatory—for example, the close adhesion which often exists between all the structures at the margin of the coloboma, and, usually, over its entire area.

On this hypothesis we can easily imagine how the inflamed vitreous pedicle (the mesoblastic intrusion into the secondary optic vesicle) would offer considerable resistance to the closure of the fissure, since it is necessary that this pedicle should be cut through before the edges could approximate closely.

Normally the closure of the foetal fissure takes place from behind forwards and from above downwards, commencing at the

optic nerve and terminating at the ciliary body, so that the *position* of the posterior edge of the coloboma may guide us in determining the *period* of intra-uterine life in which the disturbance took place, and the *size* of the affected area may indicate the *degree* of disturbance which existed in each case.

According to this theory coloboma of the optic nerve sheath indicates an earlier disturbance than coloboma of the chorioid, and the more anterior the coloboma the later the period of disturbance. Anophthalmos and extreme microphthalmos are, probably, in many cases, the result of early and extensive coloboma of the optic nerve sheath.

Coloboma of the optic nerve sheath, as seen in B. M., is an extremely rare affection, and when combined, as it is in her right eye, with a distinct and separate coloboma of the bulb, and in her left eye with a raphé-like disturbance of pigment in the line usually occupied by bulbar colobomata, it must be regarded as entirely unique.

A. Niden, of Bochum, in his article on Coloboma of the Optic Nerve Sheath in *Knapp's Archives of Ophthalmology*, December, 1879, describes four cases of the affection, in the largest of which the colobomatous disc was only three times the normal size, whereas in B. M.'s right eye the colobomatous disc is, at least, six times as large as any normal disc.

Time would fail me to dwell upon the many interesting questions which such cases as these suggest—how far the refraction of the eye is influenced by the coloboma, and how far the coloboma is affected by the primary inclination of the globe to hypermetropia or myopia. A majority of, perhaps all, colobomatous eyes are primarily hypermetropic, but many develop axial myopia from subsequent stretching of the globe backwards; or what part (an important one, I feel sure) is played by heredity; and how far such maldevelopments are associated with maldevelopments elsewhere. But before closing I may be permitted to allude to the interesting monograph lately published by Dr. Van Duyse, of Gand, on Coloboma of the Eye, and its connexion with Congenital Serous Cysts of the Orbit, in which he shows that these congenital serous cysts of the orbit, which occur either in anophthalmic or microphthalmic patients, and for which the surgeon is at times called on to operate, frequently have their origin in a coloboma, the weakened floor of which yields before the increasing intra-ocular pressure.

Considering the rarity of coloboma of the eye, it is remarkable that amongst about 500 deaf-mutes in the Dublin asylums there should be found two double cases; but, of course, the number is too small to draw any firm conclusions from regarding the connexion of coloboma and deaf-mutism. This subject, amongst others, is being worked up by Dr. Fox, for whose statistics I helped to examine the mutes in Dublin.

In the cases at present before us we have, then, a very remarkable series. Some of the conditions visible in them are such as I have failed to find mention of in any of the works on the subject which I have been able to consult.

To summarise:—(1) In M. D. we see a double coloboma of chorioid and iris of enormous size, and embracing also the optic disc. (2) In J. H. we see a double coloboma of the chorioid without implication of disc or iris. (3) In the right eye of B. M. we find an enormous (by far the largest on record) coloboma of the optic nerve sheath associated with, but separate from, a very small coloboma of the chorioid; (4) and in her left eye a smaller coloboma of the optic nerve sheath, without coloboma of either chorioid or iris, but in the position of the junction of the lips of the foetal fissure a disturbance of the pigment layer, suggesting the idea of a raphé, where the trouble just stopped short of the production of a coloboma.

From these conditions I think we may conclude:—

I. That coloboma of the chorioid may exist without corresponding coloboma of the iris, though Nettleship in his work on “Diseases of the Eye” seems to doubt this, for he says (page 166) “Coloboma of the chorioid is seldom, *if ever*, seen without coloboma of the iris, though the two are not always of proportionate size.”

II. That the closure of the fissure or furrow in the pedicle (optic nerve) takes place independently of the foetal fissure in the optic cup.

III. That the fissure may (1) close in the optic nerve and remain open in the bulb, as seen in the case of J. H.; or (2) may close in the bulb and remain open in the nerve, as in B. M.’s left eye; or (3) may remain open in both bulb and nerve, closure having taken place in the interval between the two colobomata, as in the right eye of B. M.