COLOBOMA OF THE OPTIC NERVE ENTRANCE*†

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THIS case of a coloboma of the optic nerve entrance is recorded because of the great scarcity of previous histological reports. Only very rarely is a coloboma found to be restricted to the optic nerve entrance (Duke-Elder, 1964); the great majority affect the adjacent choroid as well.

Many of the early reports concerned eyes from animals or from anencephalic stillborn infants. These as well as the instances from otherwise normal humans were reviewed by Coats (1908). By 1943 about fifty cases had been reported, most of them without histological examination (Steinberg, 1943). The first histological report of a coloboma confined within the nerve sheath of a human eye that we have been able to trace is that of von Hippel (1898), who described a cystic excavation of the optic nerve. This apparently communicated with the vitreous by a narrow neck; both cyst and neck were lined by retina. Subsequent histological studies have been reported by Rochat (1913), Calhoun (1930), Wexler and Last (1938), and Pedler (1961). Cases with illustrations of ophthalmoscopic appearances but without histological reports are those of Nieden (1879), Szili (1887), Crampton (1913), Adler (1937), Steinberg (1943), Magnus (1946), Hughes (1947), Rio Cabañas (1949), and Cashell (1951). There is probably no real difference between small colobomata of the optic nerve entrance and crater-like cavities or pits in the optic disc (reviewed by Greear, 1942).

Case Report

An infant boy was referred to Addenbrooke's hospital because of a left convergent squint. There was no family history of squint but the child's father was said to have a "lazy eye". When the child was examined under anaesthesia at the age of 21 months the left fundus was abnormal, showing a large yellowish mass at the site of the disc and extending towards the macula. This mass resembled detached retina. Vessels appeared to arise separately in this mass. The periphery of the retina appeared degenerate. The appearances were very suggestive of retinoblastoma and the eye was therefore enucleated. Since the cut end of the optic nerve appeared to be hollow, the socket was explored more deeply and another piece of tissue resembling optic nerve was removed. Post-operative recovery was uneventful. The right eye has remained perfectly normal.

Pathology

The specimen consisted of a left eye (equatorial diameter 2 cm.; antero-posterior 2.2 cm.) with a stump 0.4 cm. long of what appeared to be optic nerve. On gentle pressure, vitreous could be made to exude from a hole in the centre of the stump. Fragments of tissue from the posterior part of the orbit were also retrieved. Because the clinical findings had suggested neoplasm, the eye was

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bisected at once by a single vertical cut through the pupil and the centre of the stump. Examination of the bisected specimen (Fig. 1) confirmed that there was a canal from the vitreous to the cut end of the stump. This was lined evenly by pale grey tissue which passed forward into the globe to merge with the retina. Where it lined the canal this tissue was thicker than normal retina and the thickening persisted for variable distances into the globe, so that the site of the optic disc was surrounded by radially-arranged tapering grey ridges separated by fissures. The appearance was extremely similar to that of the fundus illustration in the report by Cashell (1951).

FIG. 1.—Bisected specimen.

After fixation in neutral buffered formaldehyde solution, one half of the eye was embedded in paraffin wax and serial sections were cut at 5μ until all trace of the optic disc had been passed; each section from the central part of the canal was stained and examined, thereafter every tenth section. The other half of the eye was embedded in nitrocellulose and serial sections were cut at 20μ until all trace of the optic disc had been passed; each section from the central part was stained and examined, thereafter every third and later every fifth section. Most of the sections were stained by haematoxylin and eosin, but some were stained by other methods, including phosphotungstic acid haematoxylin, Curtis's modification of van Gieson's stain, axon stains, and myelin stains.

Microscopical Examination.—The sections confirm that the canal at the optic disc is lined by disorganized retina. This is continuous with the retina of the globe (Fig. 2, opposite). The disorganized retina of the canal lacks rods and cones; it has poorly-developed outer and inner plexiform layers so that in some parts the outer and inner nuclear layers merge, although in other parts they are distinct. Ganglion cells are present.

The tapering grey ridges have a central core of disorganized loose gliotic tissue and often contain one or more cysts (Fig. 3, opposite). On the side nearest to the vitreous, each ridge is covered by rods and cones which are more or less normal; the base of each ridge is composed of loose gliotic tissue which rests upon the pigment epithelium. Distant from the disc the retina appears normal.

The normal pigment epithelium of the globe passes backwards to expand into a mass of collagenous tissue in which pigmented cells are numerous. This mass forms a broad collar filling in the angle between the globe and the tube of abnormal retina that lines the canal at the site of the disc. The membrane of Bruch passes backwards forming the outer boundary of this collar but becomes indistinct towards the back of the specimen, so that the posterior part of the collagenous and pigmented collar may also consist partly of choroid, vestiges of which pass backwards from the globe as far as the cut edge of the specimen.

The outermost layer of the tube is a thin collagenous sheath continuous with the sclera. The sclera and choroid of the globe are normal as also are all the structures of the anterior part of the eye.

FIG. 2.—Sections at eight different levels. Haematoxylin and eosin. $\times 5.6$.

FIG. 3.—Cysts within the ridges. Haematoxylin and eosin. $\times 40$.

Discussion

Aetiology.—Most cases of coloboma of the optic disc are associated with defects of the choroid or some other part of the eye. These are results of incomplete closure of the foetal fissure, including its posterior part. When the defect lies entirely within the nerve sheath, as in our case, the embryological defect cannot be deduced with such certainty. Some authors have favoured the view that the defect, like that in an ordinary coloboma, is of the foetal fissure but restricted to its posterior part (Steinberg, 1943). Others have suggested that the defect differs from that in an ordinary coloboma by being more primitive and representing a deformity of the primitive epithelial papilla—*i.e.* the primary optic vesicle or its stalk (Szili, 1887; Seefelder, 1927; Mann, 1957). A third possible explanation is, perhaps, that pressure on the eye from without might cause the sclera to give way so that the retina prolapsed; this mechanism was suggested by Parsons and Coats (1906) as the explanation for a cystic excavation beside the optic nerve which they found in the eye of an infant who had an orbital encephalocoele.

Clinical Presentation.—The reported adults have usually complained of poor vision since childhood. In children the condition has been discovered during routine examination for squint. In our case, like that of Pedler (1961), the presence of a mass in the neighbourhood of the optic disc led to the mistaken diagnosis of tumour. This mistake may, of course, also occur when the coloboma is not restricted to the optic disc (Buen and Fenton, 1965).

Pathology.—So few histological studies have been reported that it is difficult to generalize about the defect. The characteristic feature appears to be displacement of the lamina cribrosa backwards. In front of this displaced lamina cribrosa there may be a mass of disorganized neural or retinal tissue (Wexler and Last, 1938), or a cavity lined by retina and continuous with the vitreous. This cavity may be a small pit (Calhoun, 1930; Pedler 1961) or a large cyst (von Hippel 1898). It is unfortunate that our specimen did not include the posterior extremity of the coloboma or any trace of the lamina cribrosa, but the configuration at the nerve entrance is unquestionably similar to that in previous cases.

The eye described by Parsons and Coats (1906) differed in having the lamina cribrosa attached to the sclera at one side but detached from the other by a cystic excavation beside the optic nerve. It is not, however, certain that the disorder in that eye was identical with the group under discussion. The most extensive deformity was reported by Calhoun (1930); craters were found in both optic discs and a very large cyst in the right optic nerve. Although there was no communication between the cyst and the vitreous, it must be assumed that the colobomata and the cyst were due to the same embryological defect. Several other cases have been bilateral (Crampton, 1913; Poljak, 1928; Adler, 1937; Magnus, 1946; Hughes, 1947; Rio Cabañas, 1949). The eyes described by Rio Cabañas were also microphthalmic. In Crampton's cases the condition was familial, affecting a brother and sister. Remnants of the hyaloid artery were present in the cases reported by Rochat (1913), and Wexler and Last (1938).

Summary

A boy aged 21 months had the left eye enucleated because of suspected retinoblastoma but the abnormality proved to be a coloboma of the optic nerve entrance.

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