JUXTAPAPILLARY HAMARTOMA OF RETINA*

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THIS report is concerned with an unusual tumour-like developmental anomaly of the retina which gave rise to progressive loss of vision.

Case Report

An 11-year-old boy was referred to the clinic in June, 1959, with defective vision in the left eye, noted at a routine school medical examination. The visual acuity had been recorded as 6/6 in each eye 4 years before, and there was no history of trauma or infection, or any family history of defective vision.

Examination.—The right eye was normal with a visual acuity of 6/5.

In the left eye the visual acuity was 6/36 and could not be improved. At the posterior pole of the left eye there was an obvious oval raised greyish mass, approximately four times the area of the disc, extending from the nasal side of the disc to the temporal side of the macular area. The lesion was diffusely pigmented, with some scattered areas of darker pigment just above the disc. The edges were fairly well defined. The retinal vessels appeared normal, but their origins were partially obscured by the mass. The eye was otherwise normal. The appearance was suggestive of a lightly-pigmented choroidal melanoma. General clinical examination revealed no abnormalities elsewhere in the body and, in particular, no skin blemishes or subcutaneous nodules were present.

Progress.—The lesion was photographed (Fig. 1) and kept under observation.

FIG. 1.—Retinal photograph, showing a pigmented oval mass overlying the optic disc.

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A month later it appeared to be increasing in size and a P^{32} uptake test was performed. This showed a slightly higher uptake over the tumour in the left eye compared with the corresponding area in the right eye, but the increase was not up to the level which is usually considered significant.

Operation.—The mass continued to increase slowly in size and enucleation was carried out in September, 1959. The post-operative course was uneventful and the boy was perfectly fit 12 months later.

Pathology.—The excised eye was of normal size. After fixation it was bisected. The only abnormality present was a rounded creamy-white plaque-like thickening of the retina, 7 mm. in diameter, which overlaid the optic disc and extended laterally towards the macula (Fig. 2).

FIG. 2.—Plaque-like thickening of retina overlying optic disc. Haematoxylin and eosin. $\times 33$.

Sections of the lesion showed it to be a hamartoma lying in, and replacing, the inner layers of the retina. It was composed of abundant fibrillary glia, small thin-walled blood vessels, and melanin-pigmented epithelial cells. There was no clear-cut margin to the lesion on its deep aspect where it mingled with the adjacent normal retinal tissues. Over the greater part of the inner surface of the lesion, which abutted on the vitreous, the inner limiting membrane of the retina was replaced by a layer, one to six cells thick, of epithelial cells which were laden with melanin pigment (Fig. 3).

FIG. 3.—Pigmented epithelial cells lining surface of lesion. Haematoxylin and eosin. $\times 430$.

The degree of involvement of the retina varied considerably in different regions, but the inner layers were affected more than the outer layers. At the medial margin of the optic disc there was only gliosis of the layer of optic nerve fibres (Fig. 4). Elsewhere in the lesion there was an intimate admixture of glia, small blood vessels, and pigmented epithelial cells. A mass of this abnormal tissue completely covered the optic disc and obliterated the normal physiological cup, but deep to the lamina cribrosa the optic nerve was normal.

FIG. 4.—Medial margin of disc, showing layer of pigment epithelium and gliosis. Haematoxylin and eosin. $\times 114$.

On the lateral side of the disc there was greater distortion of the retina. Immediately adjacent to the disc the whole thickness of the retina, apart from the innermost layer of pigment epithelium, was replaced by this abnormal mass of tissue (Fig. 5, opposite). More laterally there was a progressive decrease in the number of retinal layers involved and there was no clear line of demarcation between normal and abnormal tissues (Fig. 6, overleaf).

In most parts of the lesion glial fibres predominated. The blood vessels which coursed through this glial network were irregularly arranged and varied in size up to 190μ in diameter. Many of these vessels were capillaries but some had a thin layer of muscle fibres in their walls. The melanin-laden epithelial cells, which appeared to be derived from the pigment epithelium of the retina, ensheathed the abnormal blood vessels in the lesion (Fig. 7, overleaf) and formed a lining zone on the surface in contact with the vitreous. Throughout the lesion there was no evidence of recent or old haemorrhage or of degenerative changes and only a very occasional mitotic figure was found.

FIG. 5.—Lateral side of disc, showing replacement of whole thickness of retina. Haematoxylin and eosin. $\times 114$.

Discussion

The term hamartoma was introduced by Albrecht (1904) and applied to a group of tumour-like developmental anomalies. These lesions are characterized by an abnormal mixture of the tissues that normally occur in the area affected together with an excessive growth of one or more of these tissues. They may grow during periods of growth of the body as a whole, but continued progressive growth, the hallmark of true neoplasms, does not occur. Furthermore, these lesions are benign and only rarely undergo malignant change.

On structural grounds the present lesion is clearly a hamartoma, and the clinical story is in accord—an enlarging lesion in a child with no suggestion of any traumatic or infective aetiology. Any form of hamartoma of the retina is uncommon (Reese, 1956; Russell and Rubinstein, 1959) and this lesion differs from most of the previously reported cases. It is quite unlike the predominantly glial lesions that may occur in tuberose sclerosis or von Recklinghausen's disease and, furthermore, there is no family history of similar lesions and no evidence of any anomaly elsewhere in the body. Some haemangiomas of the retina show a proliferation of glia (Carr and

FIG. 6.—Lateral part of lesion with no clear demarcation between normal and abnormal tissues. Haematoxylin and eosin. $\times 114$.

FIG. 7.—Small blood vessels surrounded by melanin-laden epithelial cells. Haematoxylin and eosin. $\times 530$.

Stallard, 1933; Cross, 1943; Souders, 1949) and this may be of a considerable degree, as in the case recorded by Wilder (1946) in which there was an intimate mixture of glia and small blood vessels giving an appearance identical with that seen in many areas of the present lesion. In none of these haemangiomas, however, has there been any proliferation of melanin-pigmented epithelial cells like that of the present case.

Under the title of "hyperplasia of the retinal pigment epithelium", Theobald, Floyd, and Kirk (1958) reported on two patients (one boy and one girl) who presented when aged 11 years with plaque-like lesions of the optic disc. In each instance vision was impaired, the lesion increased in size, and the eye was enucleated as a malignant tumour was suspected. Histological examination of the eyes showed an appearance essentially similar to that in the present case: the lesions were not neoplasms and were composed of intermingled retinal pigment epithelium, glia, and capillaries. In one case areas of hyalinization and calcification of the stroma were also present—probably due to degenerative changes. In both these patients the lesion was very like that of our present case and would be better regarded as a hamartoma than as a hyperplasia of the retinal pigment epithelium.

The clinical importance of these pigmented hamartomatous lesions lies in their close resemblance to malignant melanomas, and as yet, unfortunately, there are no diagnostic features by which they can be differentiated. It is, of course, highly desirable to distinguish between them because, although they lead to progressive impairment of vision, the hamartomas are not malignant and do not endanger life.

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