We wish to acknowledge our indebtedness to Air Commodore R. D. Gillespie and Air Commodore P. C. Livingston for putting, at our disposal their case records of a number of these patients.

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MACULAR COLOBOMA WITH BILATERAL GROUPED PIGMENTATION OF THE RETINA*

BY

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Five cases of grouped pigmentation have come under our notice in the past two years. Only one showed a coincident abnormality of the eye. It is the exception to find any lesion that might be considered congenital in the same eye as that showing grouped pigmentation. Blake, in a review of the literature until 1926 found, inferior conus, epicanthus, cataract in the fellow eye, and abnormality of the extra-ocular muscles. In none was there any visual defect. The case we propose to discuss had a macular coloboma in the right eye, and we have not been able to find a similar case in the literature. It was further unusual in the wide bilateral distribution of the pigment spots. These spots are considered to be of congenital origin. It would be legitimate to assume that the coloboma was also congenital, arising out of the same adverse factor. It is often difficult to say whether macular coloboma is congenital or not except in the familial bilateral group with skeletal deformity, or where there is some characteristic like ectasia, abnormal vessels, or coloboma of the disc to support the view. This case probably deserves record because the picture is rare and a brief consideration of the possible aetiology might be of interest.

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Macular Coloboma with Bilateral Grouped Pigmentation of the Retina

Case History

CASE 1.—R.M., Male, aged 19 years, was examined for National Service in 1943. Healthy, fair-haired youth. A truck driver with no difficulty when driving at night. V.A.R. 6/60, V.A.L. 6/6. Right eye divergent, refraction normal. The right pupil reacted less well to light directly than the left, but briskly to near and consensually. The appearance of the fundi is as indicated in the diagrams (Fig.). The right eye showed a roughly circular coloboma, much larger than the disc with a non-pigmented base and a pigmented border. The sclera was not ectatic and retinal vessels passed over the surface of the defect smoothly. The disc was normal and identical with its fellow in colour. The eye grounds presented grouped pigmentation of the retina, and in each eye, in all four quadrants of the retina were groups of spots. The sectors, or wedges of spots followed the main retinal vessels.

CASE 1.—Diagrams of Fundi.
very closely and the shape of the dots varied from small black dots near the disc to larger and paler, crescentic, round, or polyhedral patches which tended to fit into one another, nearer the periphery. The base of the wedge did not extend to the equator. The visual field was of normal extent in each eye and no relative defect was discovered within the field in the left eye. The right eye had an absolute central scotoma. The blood Wassermann reaction was negative. No abnormal pigmentation of the iris, or conjunctiva was observed.

Discussion

The frequency of misplacement of tissues in abnormal eyes was noted by Coats (1909). The explanation later adduced was that the genic constitution was upset. These ectopic tissues were somatic mutations. Parsons found histologically that "bear's paw" spots consisted of intensely pigmented cells and glial-like
tissue, some of the cells having migrated into the retina. But pigment migration is a common feature of retinal pathology. It is the fundus picture that makes us think the condition is of prenatal origin. Nothing like it arises out of an observed post-natal retinal disease process. Most authors agree that the spots do not change throughout life. They are larger and paler near the equator and the group of spots is consistently wedge shaped with the apex near the posterior pole. These facts would be explained if the globe expanded most at the equator during the last weeks of gestation. Miss Mann has been good enough to inform us in correspondence that she considers that this is the case. So that, given a group of spots near the posterior pole in the sixth month they might acquire the character and distribution of grouped pigmentation by term on a simple mensuration basis.

It is clear from the evidence of experiment in tissue culture that varied environmental conditions may produce the same type of derangement provided the disturbance factor is active at the same stage in development. The time is important as well as the agent. Sporadic congenital macular coloboma, or dysplasia may be regarded as a developmental failure or as the result of foetal inflammation. Miss Mann considers that the facts are best explained on the inflammatory concept. She further states that the characters of the coloboma may denote the time of origin of the lesion; coloboma with abnormal vessels occurring at the time when the vitreous is full of vessels in the 3rd month, non-pigmented coloboma with ectasia occurring at the 6th month, and pigmented coloboma in the later months. The last is nearest to the adult disease pattern. Any environmental adverse factor, inflammatory, hormonal or physical acting in the early months to produce defect, results in gross deformities which are roughly similar in plan but have no resemblance to the aspects of adult disease. Late foetal choroiditis may be indistinguishable, microscopically and histologically from adult choroiditis. In the foetus the macula suffers more readily because it is at the top of the axial gradient after the closure of the optic cup. In this connection the papers of Vail, Koch, Wolf, Cowan and Paige and others in central choroiditis of infants infected by toxoplasma are interesting. Many infants of the observed series were considered to have been infected in the later weeks of gestation. Complement fixation and toxicity tests for toxoplasma gave a large percentage of positive findings in affected children, and many of the mothers were also positive (Sabin, Sabin and Buchman, Warren and Sabin). Vail says, "It is significant that in nine out of ten cases in which the sera contained neutralising bodies for toxoplasma the only clinical manifestation was that of a chorio-retinitis of unknown etiology with a predilection for the macular region."
So that we may regard our case as a developmental, or inherent fault, showing arrest and aberration; macular arrest and abnormal development of pigment as a somatic mutation. We know little of the laws that govern mutation. Experimentally the gene constitution can be altered by X-rays in Drosophila to produce white facets in the eyes of an insect which would otherwise be uniformly red. The toxins of disease may on occasion be the initiators of somatic mutations in actively differentiating tissues. Alternatively, one may simply regard this case as a foetal choroiditis, having the main characters of any patch of old choroiditis, namely, atrophy at the centre of the lesion and pigment proliferation at the periphery, the pattern of the peripheral pigmentation being subsequently altered by growth of the globe. The grouped pigmentation was unusually well-marked being present in all quadrants of both eyes. It might be suggested that a foetal inflammation had destroyed the macula and given rise to the surrounding pigmented changes, these changes assuming the characteristics of grouped pigmentation with further growth of the globe.

REFERENCES


SECONDARY CARCINOMA IN THE ANTERIOR CHAMBER*

BY

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Secondary carcinomatous deposits within the eye probably occur with greater frequency than the literature would lead one to suppose. Stallard (1933) gave a ratio of 1 : 140,000 in the Moorfields records. There is little doubt that if routine examinations were

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