Tapeto-Choroidal Degeneration (Choroideremia)

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Shortly after the Meeting of the Netherlands Ophth. Society, in December 1958, when Pameyer, Waardenburg and I discussed a family with choroideremia, another case of this affection was seen in its incipient stadium thanks to the collaboration between the Eye Clinics of Rotterdam and Utrecht. Since there was no advanced case of it in the family under discussion, the diagnosis could probably not have been made without further ado of the family described earlier.

As particularities at the early stage of the choroideremia I would like to point out:

The primary affection of the tapetum, with pigment migration only in the deepest layers of the retina (thus not, as in tapeto-retinal degenerations, rising to the superficial layers of the retina); the secondary atrophy of the choroid (simple disappearance without preceding sclerosis of the vessels), with subsequent atrophy of the retina;

the great similarity of the picture of the carrier (female transmitters) with that of the choroideremia at the beginning stage.

The 8-year-old patient complained of hemeralopia and slightly diminished vision¹. Optic disc and retina: practically normal, except a slight peri-papillary atrophy. Macula: a speckled appearance, as though it had been strewn with black pepper. An identical picture in both eyes. Proceedings towards the equator: coarsely-meshed structures, whereby both the choroid and the retina are beginning to atrophy in the meshes. Consequently the meshes are elevated at the periphery: not a normal retina (as in atrophica gyrata retinae et chorioideae) but a definitely untidy picture. The carriers—the mother and two aunts— show very typical abnormalities in fundo. Optic disc: normal. No peripapillary atrophy. Macula: untidy; decidedly too much pigment but not the fine-dust design seen in the son. Periphery: more and more disorderly as we approach the equator. Past it, the same typical meshes laden with pigment that we know from the affected boy.

¹ Will be published in the British Journal of Ophthalmology.

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Heredity is X-chromosomal, as appears from the pedigree chart of this new family. The patient will beget only normal sons and carrier daughters. The transmitter will have 50% affected children, either patients (sons; 25%) or again carrier daughters (also 25%).

Regarding the visual functions the following: Our case shows reduced retinal sensitivity and an enlargement of the blind spots, in the Goldmann perimeter. In the carrier examined (the mother): no clearly reduced retinal sensitivity. Dark adaptation is seriously disturbed, with progressive deterioration in the patient. Probably there is some disturbance in the rate of dark adaptation in the carrier examined, which is not in the least unlikely considering the serious fundal changes. The ERG of the patient shows absence of scotopic responses and serious impairment of the activity of the cone mechanism. No abnormalities in the carrier was found.

It is hoped that this presentation has been able to give a better understanding of the picture of choroideremia in its incipient stage, at which, unless there is a hint from a more advanced case,

diagnosis can be particularly difficult, but information about which is so important in connection with the heredity of the affection.

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Once again the Oculo-Cardial Reflex

By J. TH. PLANTEN

In a former report electro-cardiographic findings were recorded as resulting from the oculo-cardial reflex during strabismus operations (Planten, 1958).

In the following case history this reflex played an important role in our consultations.

Miss A.

15 years old

was hit in the eye from a short distance by a bullet from an air-gun. When she was taken to the hospital

X-rays were first made. During this time the patient turned pale and became clammy and began to vomit. Her pulse was normal

light irregular

44 beats a minute. The X-ray photos showed that the bullet was not lodged intracranially (Fig. 1). The clinical condition of the girl

however

led to the question of whether the central nervous system had been injured.

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