Choroidal Atrophy

Clinical and Genetic Types

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In ophthalmology, as in medicine generally, there has been a rapid development of treatment to combat infections and allergic conditions, but the choice of therapy for degenerative, hereditary conditions is—admittedly—just as small as before. The proportionate role played by the genetic degenerative eye lesions as causative factors of blindness, or impaired vision, has thus greatly increased.

Related to the large and not very well-defined group of degenerative eye lesions called the tapeto-retinal degenerations, there is a somewhat disputed group termed choroidal sclerosis, or perhaps more correctly, choroidal atrophy. As mentioned later in this paper, there is histological evidence of choroidal atrophy rather than sclerosis. The term 'atrophy' will therefore be preferred in this article.

According to Sorsby (1939), there are three clinical forms of choroidal atrophy, first, *central areolar choroidal atrophy*, first established by Sorsby (1939) as a clinical entity, secondly, the *diffuse choroidal* form, described by Morton (1893), Frost (1896), and Harman (1902), and lastly *peripapillary choroidal atrophy*, described by Haab (1895), Harman (1902), Cuperus (1903), and Di Marzio (1938).

Central areolar choroidal atrophy is characterized by the development in both fundi of a sharply circumscribed central oval area of choroidal atrophy with disappearance of the choriocapillaris and the retinal pigment (Fig. 1). Retinal vessels can be seen traversing the atrophic zone. The size of the atrophic area is about 3 to 4 times that of the disc. The earliest ophthalmoscopic appearances are evident between the ages of 20 and 40, and are of an exudative oedematous type somewhat resembling that of macular degeneration. The development of the disease is slow and gradual. The end-stage is reached at about the age of 50–60 years. Visual acuity is then usually reduced to less than 0.1, while

the peripheral fields are normal. There is no night-blindness.

Several sporadic cases have been published, first, by Nettleship (1884), who noted the condition in a 60-year-old woman, and later by Retze (1901) and Thompson (1905). Sorsby (1935, 1939) was the first to draw attention to the genetic nature of the disease, noting the occurrence of the condition in 2 brothers, while Waardenburg (1952) observed the condition in 2 sisters, and Sorsby and Crick (1953) in 12 other cases in 4 families. Sandvig (1955, 1959) reported a pedigree (Fig. 2) with 13 affected members, 6 men and 7 women, through 4 successive generations, giving conclusive evidence of an autosomal dominant mode of inheritance. Thus, one is justified in stating that the inheritance is autosomal, either recessive or dominant, with no predilection for either sex.

FIG. 1. A typical picture of central areolar choroidal atrophy.

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FIG. 2. Pedigree.

Diffuse generalized choroidal atrophy is characterized by generalized atrophic changes throughout the fundus, where the larger choroidal vessels stand out prominently as a greyish-white network. The age of onset generally lies between 20 and 30 years. This type also begins in the foveal and perifoveal area, but soon involves the entire choroid. Consequently, central scotoma develops at an early age. The periphery of the visual field remains undamaged for a long time. Since hemeralopia is usually absent and the retinal vessels and disc are normal, it is unlikely that the process is primarily a retinal one. The choroidal vessels are converted into white streaks. Later on, diffuse atrophy appears, associated with irregular dystrophy of the tapetum nigrum and pigmentary accumulation, most frequently localized in the centre and periphery.

The condition was first described by Morton (1893). The mode of transmission is usually autosomal dominant, as shown by Tobler-Berg (1937: 8 cases in 3 generations), François (1949: 6 cases in 3 generations), and Sorsby and Davey (1955: 4 cases in 2 generations). According to Waardenburg, Franceschetti, and Klein (1961), the family described by Tobler-Berg (1937) does not belong here, because there was an atypical retinal dystrophy with precocious affection of the foveal region and development of sclerosis and atrophy of the choroid.

In other cases, however, the familial appearance of the disease without traces backwards or forwards, suggests a recessive mode of inheritance (Sorsby 1939: 3 sibs out of 8). Lastly, sex-linked transmission has been recorded by several authors. There is, however, some evidence that such cases are not true cases of diffuse choroidal atrophy, because they

tend to progress to an end-stage of total atrophy very much resembling that of choroideraemia.

In peripapillary choroidal atrophy, the pathological changes with exposure of the white or grey vessels usually start around the disc, spreading later on to involve the posterior pole as a sort of central choroidal atrophy. However, in contrast to 'genuine' central areolar choroidal atrophy, the lesions have no clear-cut margins. In addition to the solitary cases reported, there are familial cases on record, for example Sorsby (1939: 2 sisters) and Waardenburg (1952: 2 sisters, with 4 normal sibs) pointing to an autosomal recessive mode of inheritance.

As stated briefly in the opening paragraphs of this article, there is a discrepancy between the clinical and histological pictures in this condition. While the clinical picture is one of vascular sclerosis, histological examination (however scanty) reveals complete disappearance of the choriocapillaris, of the pigment layer, and of the neuroepithelium in the centre; there are also secondary irregularities, e.g. ruptures, and lamellar structure of Bruch's membrane, and only very slight sclerosis of the large choroidal vessels. In the opinion of Babel (1958), the clinical picture of angiosclerosis may be produced by a sharper contrast of the vessels to the atrophic zone (Ashton, 1953; Babel, 1958; Howard and Wolf, 1964).

Histopathological study of central areolar choroidal atrophy has been greatly neglected. More extensive studies are necessary before we can emerge from the present stage of theories and speculation regarding the pathology and pathogenesis of choroidal atrophy or sclerosis.

It is difficult to say whether choroidal atrophy represents a clinical entity. The primary fault seems to lie in the choroid, the changes in the pigmentary epithelium and retina being secondary. The fact that choroidal atrophy has been associated with other tapeto-retinal degenerations probably indicates some sort of relation. According to François (1958), the classification of heredofamilial degenerations of the chorioretina is too artificial, being based on the conception of a great number of autonomous affections, characterized on the basis of morphology, age, and localization. There are so many intermediary forms between all these varieties of heredofamilial degeneration that the idea of a unique heredodegeneration comes naturally, especially since transitional forms and apparently different types may be found in the same family.

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