CHOROIDEREMIA AND GYRATE ATROPHY
OF THE CHOROID AND RETINA

by

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With 37 figures, 16 photographs and 5 tables

Introduction*

Until recently, choroideremia (progressive tapetochoroidal degeneration; see page 41) and gyrate atrophy of the choroid and retina were regarded as uncommon hereditary ocular affections. Only since 1942 have some publications on choroideremia appeared in the Netherlands, and reports on gyrate atrophy of the choroid and retina have been largely confined to casuistics.

Very little is known as yet about the clinical picture of the early stages of both these affections. Consequently they are frequently confused with each other or with (fundus) anomalies such as choroidal sclerosis and tapetoretinal degeneration.

The object of this study was:

1. To trace as many cases as possible in order to gain an impression of the incidence of choroideremia and gyrate atrophy of the choroid and retina in the Netherlands. In addition, to enlarge our knowledge of the clinical picture of these two conditions, especially in their early stages, by an exhaustive and exact investigation.
2. To make an objective analysis of the genetic aspects of these two clinical entities.

The study was carried out with the support of the Netherlands General Association for Prevention of Blindness which, together with the ophthalmological clinics and many ophthalmologists, co-operated in collecting the material. The genealogical research was done by Ir. T. W. SIERTSEMA, the Association’s genealogist.

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PART I

CHOROIDEREMIA

Brief historical review

MAUTHNER was the first (1872) to describe the clinical picture, which he designated "choroideremia" because he interpreted the findings as congenital absence of the choroid. It was later found that the disease is progressive, and it is still dubious whether it can be congenital. SORSBY et al. (1952) consequently referred to "progressive choroidal atrophy", while WAARDENBURG (1958) and PAMEIJER et al. (1960) described the condition as "progressive tapetochoroidal dystrophy". The motives behind these designations will be discussed later (page 40).

MAUTHNER's report was followed by descriptions of a large number of isolated cases, all encountered in males, by such authors as COWGILL (1892), LANDMAN (1906), MARBAIX (1908), SMITH & USHER (1916; cf. RIDDELL 1933-1934), CONNOR (1919), ZORN-SCHUTZBACH (1920-1938), BECKERSHAUS (1926), DIMMER & PILLAT (1927), PARKER & FRALICK (1931), DE SCHWEINITZ (1931), BAHN (1932), BHADURI (1934), WILMER (1934), BENEDICT (1937), WÜRDEMANN (1937), DI MARZIO (1937), FRIEDMAN (1940), MAGDER (1945), MEYRAN (1948) and others.

Familial occurrence of the condition in brothers was described by KÖNIG (1874), BULLAR (1898), THOMPSON-WARDALE (1899-1906), ALEXANDER (1910), WOLF (1930), WERKLE (1931), BEDEL (1937), BENCI (1938), SORSBY (1939; originally reported as choroidal sclerosis), SCOBEE (1943) and ESTERMAN (1947). A number of these reports were based on anamnestic data.

The confusion with other hereditary affections of the choroid and retina was ended in 1942, when two Dutch ophthalmologists - GOEBLOED and WAARDENBURG - demonstrated in independent studies that choroideremia is subject to intermediate X-chromosomal transmission. This discovery greatly contributed to a more accurate definition of the place of choroideremia in the group of choroidal and retinal dystrophies. GOEBLOED reached his conclusion on the basis of personal observations in a family including one patient and two women showing only slight fundus changes, and consequently considered to be carriers of the mutant gene, and also on the basis of an analysis of the pedigree of ZORN-SCHUTZBACH. WAARDENBURG formed his conclusion on the basis of a penetrating analysis of the literature, after having had occasion to re-examine SMITH & USHER's patient in 1939. GOEBLOED and WAARDENBURG based their postulate upon, among other things, the fact that males show the complete clinical