Tapetochoroidal Degeneration

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Summary

An investigation into the occurrence of choroideremia has been carried out with the help of the Netherlands General Association for the Prevention of Blindness.

Forty-five affected men and 62 carriers were found. The majority of these patients underwent a detailed examination in the clinic.

The fundus pictures of the affected men and also those of the carriers vary greatly, and a wrong diagnosis can easily be made without the help of a pedigree investigation into the occurrence of other diseases, such as:

atrophia gyrata chorioideae et retinae, choroidal sclerosis, atypical tapeto-retinal degeneration.

Fundus pictures at different stages of choroideremia will be shown, and also fundus pictures of 3 carriers, suffering from subjective and objective defects.

(Will be published in extenso in Documenta Ophthalmologica.)

Discussion

Waardenburg: Your very extensive investigation has revealed that a heter-ozygote female carrier can undergo the same process as the male hemizygote, on the strength of the fact that the normal gene, which is absent in the male but present in the carrier, does not work in the latter. This reversal of the dominance, which has been observed occasionally in other X-chromosomal anomalies, e.g., in tapeto-retinal dystrophy and eye albinism, can thus here also lead to a similar breaking-down process as in males.

This does not, however, alter the fact that we must as a general rule, in my opinion, look for the primary cause of the condition in the tapetum nigrum, which, on binocular fundus examination of carriers and early male

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cases, is clearly seen to resemble a rasp with thicker and thinner areas. In male patients the areas soon run together, to be followed by gradual disappearance of the choroid; in carriers the normal gene usually appears to hinder this process or even to prevent it altogether.

The best nomenclature, instead of choroideremia which implies absence at birth, is still progressive tapeto-choroidal dystrophy.

De Haas (Rotterdam): In their recent report delivered to the Société Française d'Ophtalmologie Franceschetti, Francois and Babel say that choroideremia, X-chromosomal hereditary choroidal sclerosis and X-chromosomal hereditary tapeto-retinal degeneration cannot be differentiated genetically, and that they have only dealt with them separately for didactic reasons. Does the speaker agree with this on the grounds of his own observations?

Kurstjens: In choroideremia a fundus picture is often found which closely resembles that of choroidal sclerosis, so that choroideremia and X-chromosomal hereditary choroidal sclerosis are difficult to tell apart.

Further support for this hypothesis is that in one choroideremia pedigree a woman was found with a tapetum-like reflex in the fundus, similar to that described by I. Mann.

In heterozygotes of the X-chromosomal hereditary tapeto-retinal degeneration a tapetum-like reflex has been observed more frequently.

To what extent

however

this tapetum-like reflex may be taken as a sign of the carrier state in choroideremia cannot be decided in this one case

especially as no abnormalities were observed among this woman's descendants.