Atrophia retinae pigmentosa atypica

G.J.M.E.N.

Timmerman

Michielsgestel

Author's address: Dr. G. J. M. E. N. Timmerman, Papaverstraat 37, St. Michielsgestel (The Netherlands)

A family is described that consists of 12 persons suffering from tapetoretinal degeneration. The fundus picture of the father appears, at first glance, to be completely different from that of the children; study of the literature, however, shows that the condition fits the description of atrophía pigmentosa atypica, with, as sub-groups, the multi-pigment and the pauci-pigment forms.

The youngest child of the family is the only one who appears to be completely normal on extensive examination (electroretinography, electro-oculography, dark adaptation curve, colour-vision investigation).

The EOG is the most frequently abnormal feature. This can be explained by the localization of the pathological changes in the retina and choroid. The type of heredity is not known with certainty; recessive auto-somal is most probable. Theoretically, the mother can be a carrier. Consanguinity is not a sine qua non.

Discussion

Mrs. Schappert-Kimmijser: This paper demonstrates once more the need of a follow-up especially in the case of tapetoretinal dystrophy. For this purpose it is necessary that the ophthalmologists and particularly the eye hospitals should register all cases of tapetoretinal dystrophy.

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