A Diagnostic Difficulty Solved by Co-Operative Effort

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During an investigation of a large number of children from institutions for the partially sighted and blind in the Netherlands initiated by the Netherlands General Association for Prevention of Blindness it turned out that the diagnosis of tapetoretinal dystrophy in two brothers, made by three oculists on the basis of poor vision, nystagmus and fundus picture, became questionable when a normal ERG was found.

Information about the family brought in by the social worker of the Institution led to the gathering of information about the eye affections of three distant cousins who also had very poor eyesight.

On the basis of particulars furnished by the consulting ophthalmologist (poor vision, nystagmus and retinal atrophy), it was possible to conclude with almost complete certainty that they were suffering from the same disease.

The provisional pedigree then drawn up pointed in the direction of X-chromosomal heredity, after which, by elimination, ocular albinism could reasonably be suspected. Further investigation confirmed this assumption: both in the patients and the carrier (mother) the iris proved to be diaphanous. Further enquiry in the family led to the drawing up of a more complete pedigree; this revealed many affected persons and carriers.

The importance of a correct diagnosis to the family is pointed out (in this case the knowledge that the condition does not progress and that certain branches of the family will certainly be unaffected) and emphasis is laid on the value of the ERG, the drawing up of a pedigree and the co-operation between various specialists.

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Henkes, demonstrating several fundus pictures, wishes to point out once again that pigment-anomalies occur in the fundi of carriers of ocular albinism. For this reason it is easy to understand that in these families a mistaken diagnosis of tapetoretinal degeneration is frequently made.

Waardenburg: For the sake of clarity it seems to me desirable to draw the attention of colleagues who have never diagnosed it to the fact that in ocular albinism the stroma of the iris may contain some pigment and the eyebrows and hair may also show a certain amount of pigmentation. Thus besides complete and incomplete universal albinism

which is recessively hereditary

recessive X-chromosomal ocular albinism also exists. In this last condition the carriers always have a diaphanous iris.