Entirely normal parents had a child which at the age of 6 months was presumed to be blind. We saw the boy then for the first time; he showed nystagmoid movements which sometimes developed into real nystagmus. The pupils did not react to light, there were no following movements to light, but the fundi were completely normal. The boy is now 4½ years old and the condition is unchanged. He is very intelligent. The paediatrician found one peculiarity, viz. a typical herringbone pattern of the nails.

Ten months ago a sister was born who is also blind, with normal fundi. The shape of her skull is somewhat peculiar (brachy-cephaly) with a “dip” in the occiput. She has the same nail pattern. ERG and EEG were carried out under slight anaesthesia. The boy showed diffuse EEG disturbances, especially occipitally. In the girl there were marked EEG disorders. In both children, the ERG was absent.

Summarizing, two successive children in one family who are blind with normal fundi, absent ERG, disturbed EEG are presented. There are no further children. What kind of affection may this be?

Is this the disease discovered by Alström on the basis of a statistical genetic investigation: heredo-retinopathia congenitalis monohybrida recessiva autosomalis? It is clear that identification of the anomaly in these children with that described by Alström can only be based on a genetic investigation.

There is no consanguinity between the parents as far as we know. They have both a bilateral visual acuity of 1, normal fundi and normal visual fields. The father is nightblind; he is the hemeralopic patient of one other report. More important detail is that a child of the mother’s sister is, or has been, treated in the Institute for the Blind at Grave. A brother of the mother’s mother was stated to have had a blind child that died at the age of 1 year, it is evidently difficult to draw conclusions from these data.

Notwithstanding appreciation of the genetician’s work, the clinician has to answer the question as to the cause of the blindness. Does the absence of the ERG justify the presumption that the cause lies in the retina? Waardenburg seems to have assumed this in an article read before this Society in Groningen and entitled: “Does Agenesis of Dysgenesis Neuro-Epithelialis Retinae Exist, either Associated with Keratoglobus or without it?”. Mrs. Schap-pert followed suit in a report read at Rotterdam, and mentioned tapetoretinal degeneration. At present she calls it tapetoretinal dysfunction. What then, however, should be said of the histology of an eye of one of Waardenburg’s patients? This patient had been blind since birth and had a normal fundus in his right eye, whereas that of his left eye was not visible owing to corneal and lenticular opacities. The left eye was painful and had to be removed. Histological examination revealed
signs of long-standing glaucoma. However, the rods and cones were intact. The argument could now be raised that the histological picture may seem normal, but that these cells evidently do not function. In that patient the ERG was also absent. The question may be raised whether normal rods and cones have been found in other cases of absence of ERG. In this connection it is interesting to note that Zetterström found that the ERG cannot be elicited in newborns. This is also known of new-born animals.

It may be asked whether it is possible for a patient who has been blind since birth to have an ERG, but the answer cannot yet be given. Another factor is involved in young dogs with hereditary retinal degeneration described by Parry, Tansley and Thomson. In these, a retinal potential was initially present, but it disappeared later, with the development of retinal degeneration.

At the request of Vierhout and Waardenbmg we collected 3 other children from one family, among whom uniovular twins, all born blind. An older daughter from this family is normal. In these children the EEG, as far as it has been taken, is normal. The ERG is absent. We would not call the fundi entirely normal, especially not in the eldest boy. In the periphery there are dark and white spots, whereas the centre is normal. However, we should like to close this article with the words used by Zeeman in his Textbook of Ophthalmology, Part II, when dealing with ophthalmoscopic examination. He says: “For evaluation of the fundus neither the total impression of the entire picture, nor a comparison with pictures seen earlier or with illustrations in an atlas is sufficient. Even pictures that have never been seen before have to be interpreted ...” This is the difficulty in these cases.

Discussion.

Leffertstra asked: How can in practice a differentiation be made between the dysplasia retinae described and cases of retinitis pigmentosa sine pigmento? In retinal dysplasia i’undal changes may also develop.

Zeeman, W.P.C: Ophthalologica 137: 425–426 (1959)

Report of a Study of Dr.G.D. Hemmes on the Problem whether the Prognosis of Retinoblastoma in the Netherlands can be Favourably Influenced.

By W. P. C. ZEEMAN, Amsterdam.

Some time ago we've suggested the idea of early treatment of retinoblastoma by means of systematic follow-up of the children of retinoblastoma patients and their close relatives. The Queen Wilhelmina Funds kindly allowed a grant to the Netherlands General Society for the Prevention of Blindness for an orienting study of the practicability of such a scheme and of the factors which inhibit early diagnosis. G. D. Hemmes has carried out this study and laid down the results in a report.