A Practical Aid to the Recognition of Leber's Disease

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It is with great pleasure that I draw your attention to a service offered by the Netherlands General Association for the Prevention of Blindness, which is, as far as I know, unique in the world. A number of eye diseases are difficult to diagnose, and among these the hereditary eye diseases are on the increase. The reason for this is, on the one hand, that our therapeutic and diagnostic arsenal against non-hereditary disease is growing, and on the other hand, that excellent welfare increases the possibility that hereditarily-disposed persons will raise large families with the hereditary trait. Viewed in this light, the practice of bringing together patients of both sexes suffering from the same (hereditary) diseases in institutions and associations is not without danger.

As the hereditary character of these diseases is an important diagnostic criterion, this aspect must be thoroughly investigated. In a normal medical practice there is no time or opportunity for performing these time-consuming investigations.

As a result of the investigations into Leber's disease in the Netherlands, archives have been compiled covering all the relatives of Leber patients discovered in this country who, according to the special manner of transmission, have a chance of developing the condition.

The desirability of such archives was stated by Waardenburg as early as 1924.

These archives are very helpful for the establishment of the diagnosis. Leber's disease is not specific in its symptoms. In the 3 years in which these archives exist, a probable diagnosis of Leber's disease has been made 16 times by discovering a relationship in the female line to sufferers from the disease.

These archives are at the disposal of all ophthalmologists and can be consulted by application to my address. Please will you

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supply the surname, Christian names, date and place of birth, not only of the patient concerned but also of the mother and mother's mother. This is of importance for tracing possible X-chromosomal inheritance. Hereditary transmission by the male has not as yet been observed, but if such a case should present itself it could be discovered by means of these archives. In a case in which a woman and her children were omitted in a letter from the municipal registration authority, the patient could be localized at once, through the name of the grandmother, as belonging to one of the known pedigrees.

In return for the information given, I should like to ask for the clinical details of the cases, which are of value for the archives.

I hope that the practical result of this investigation will be the stimulation of all ophthalmologists to co-operation in the other investigations undertaken by the Netherlands General Association for

Prevention of Blindness, so that the amount of information which can facilitate the diagnosis in difficult cases will be increased.

References

van Sensus, A. H. C.: Leber's disease in the Netherlands. Docum. Ophth. 17: 1-162 (1963). Waardenburg, P. J.: Beitrag zur Vererbung der familiären Sehnervenatrophie