Hereditary Vitreoretinal Degenerations

Developments in Ophthalmology

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Hereditary Vitreoretinal Degenerations

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The problem of achieving differential diagnosis in cases of vitreoretinal degeneration can challenge even the most experienced scientist. In the course of degenerative diseases, various changes in the vitreous body and retina combine with a high degree of variability, often making it hard to set the boundary between normal and pathological conditions, to attribute findings to specific syndromes, or to distinguish between hereditary and exogenously triggered disorders. Without a doubt, the improvement of subjective functional tests, new possibilities in morphological diagnostics due to angiofluorography, and the supplementation of clinical findings with electrophysiological studies have markedly enhanced diagnostic procedures in the last 10 years. This, however, does not disguise the fact that many individual observations remain inadequately explained. In this monograph, Walter Lisch has pointed out that further improvements can be made in the diagnosis of hereditary vitreoretinal degeneration using a method in which morphological findings of characteristic clinical signs are subjected to precise analysis. Proceeding from individual observations of our out-patients with disease of the retina, he was able to detect a large number of families presenting the different progressive forms of vitreoretinal degeneration, establish the exact diagnosis through genetic determination, and then analyse the morphological findings of various characteristic signs observed in the large patient sample. The resulting
morphological picture permitted the elaboration of new criteria for differentiating the various stages in the evolution of hereditary vitreoretinal degeneration and for distinguishing these findings from non-hereditary vitreoretinal diseases. In addition, the family studies, which included observations of juvenile probands, made it possible to precisely detect and describe the initial stages of the disease and to set the boundary between these findings and variants of the normal state. Studies focusing on the evolution of the disease, as well as the examination of elderly patients, produced new information on the final stages of various hereditary disorders. Surprisingly, genetic studies are not only possible in less populated and demographically isolated areas, but also in large, densely populated regions such as the Ruhrgebiet. Walter Lisch has already proved this in connection with hereditary diseases of the cornea and fovea.

Foreword VIII

In one work, Walter Lisch compiles the analytical studies which we have followed with great interest for years. The individual results of these studies have been stimulating and beneficial for clinical work. We are certain that this publication will be a starting point for further investigations in this field, which is so important to ophthalmology.

Together with Walter Lisch, we would like to thank Professor Blodi, Iowa City, for personally undertaking the task of translating the manuscript into English.

Dortmund, June 1982

Klaus Ullerich