WENT, VERMEIJ - KEERS, VAN DER LINDEN

EARLY DIAGNOSIS AND PREVENTION OF GENETIC DISEASES

BOERHAAVE SERIES Nr. 11

EARLY DIAGNOSIS AND PREVENTION OF GENETIC DISEASES

EDITED BY

L. N. WENT, D.SC., CHR. VERMEIJ-KEERS, M.D., AND
A. G. J. M. VAN DER LINDEN, PH.D.

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EARLY DIAGNOSIS AND PREVENTION OF GENETIC DISEASES

PREFACE

The present volume of the Boerhaave series intends to be a reflexion of our present knowledge in the expanding field of early diagnosis and prevention of inherited disorders.

Early diagnosis can mean detection of a carrier state in one or both potential parents, detection of a specific inherited disease in a previously born child or during a pregnancy at risk, or during various stages of the life of an already born individual.

The first chapters will discuss inherited disorders manifesting later in life. In Huntington's Chorea detection may be possible from the age of 20 or 30 onwards, while in myotonic dystrophy it will be seen that detection may be possible soon after birth.

Retinoblastoma serves as example of a disease manifesting in early infancy and which is partially treatable by surgical intervention. The same early onset does occur in phenylketonuria and cystic fibrosis. Both diseases provide examples of the possibilities of population screening, while in phenylketonuria an eminently succesful treatment is found in a restricted diet.

For the usually early manifesting sphingolipidoses no treatment exists as yet, but enzyme treatment might in the near future become a reality.

Spina bifida and anencephaly are not simple inherited Mendelian disorders but they are discussed in this volume because prevention is possible by antenatal diagnosis and subsequent interruption of the pregnancy if requested by the future parents. Further views on antenatal diagnosis notably in inborn errors of metabolism are presented in a following chapter.

Haemophilia is discussed as an example of a sex-linked disorder in which detection of heterozygote females is feasible in many instances.

In an overall picture of the problems and possibilities around family detection of inherited diseases some of the aspects mentioned in preceding chapters are summarised.

The difficulties and future prospects of genetic counseling and screening at the population level receive ample attention and are discussed in the second part of the book.

The last part is dedicated to ethics. It begins with explanations by the ethicists how one arrives at moral decisions and who is involved in reaching these decisions; this is followed by a general discussion.

The editors

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