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Genetic Counseling

A Guide for the Practicing Physician

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With 27 Figures

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Preface

“An ounce of prevention is worth a pound of cure”

In medicine the truth of this statement is so self-evident that it is simply taken for granted; and yet it has become mere lip-service for many a doctor, since his work is almost exclusively concerned with the treatment of those who are already ill. This applies not only to the treatment of patients but even more to that of entire families. Many doctors are as yet unaware that the appearance of serious, sometimes fatal diseases can be avoided by preventing the conception of sick human beings. Our knowledge of genetics permits the relatively accurate prediction, based on statistical probability, of the recurrence of genetic defects (anomalies) and diseases within families.

Our patients are frequently aware that such predictions are possible. In an effort to prevent the birth of defective children they try to inform themselves. However, in the practice of the individual doctor this sort of inquiry does not occur with such frequency that he is forced to concern himself systematically with these problems. Should he be confronted with such an inquiry, the doctor conscientiously tries to recall what he has read—once upon a time. In many cases the doctor's actual contact with genetics has occurred so long ago that he has forgotten most of the particulars, or genetics was never taught when he was at university. Consequently, the general practitioner usually has no exact knowledge on which to base his advice. Either he evades the problem with vague generalities, or he remembers various statements about “heredity” that almost always lead to false conclusions.

From discussions with our colleagues, we know that they recognize the problems and worry about them, but simply do not have the time to thoroughly study the highly specialized genetic literature available.

This book is an attempt to fill this void. We have made an effort to keep it as short and clear as possible and to limit it to the impor-

tant and most frequent genetic abnormalities. In particular, we have tried to take into consideration the difficulties of the average student in understanding genetic logic and to eliminate the most common errors.

This guide is not designed to provide more than basic information. No reader will arise from the study of this volume as an expert genetic counselor. That requires, in this as in all other sciences, knowledge of the highly specialized literature as well as extensive experience. Some geneticists therefore take the position that the general practitioner (or specialist in any other field of medicine) cannot possibly give proper genetic counsel to his patients. Because he is not a genetics expert, he should, without exception, refer all such cases to the geneticist. This point of view would condemn this guide as potentially more harmful than helpful in that it might increase the cases of well-meaning error as well as encouraging those who are not competent in this field to deal with problems which are beyond their capacity.

We, obviously, do not share this pessimistic standpoint. In our opinion, there are cases in which an interested and informed general practitioner can give fully adequate counsel. In these cases it is of great advantage to the effectiveness of the advice that family doctor who has the confidence of his patient(s) also carry out the counseling. Opposed to these clear cases are the highly complex ones which exceed this guide's limitations. In such cases the doctor must recognize the limits of his knowledge and consult a genetic counseling clinic or a specialist. Exactly where the limits are to be found in a specific case must be left to the conscience of the individual doctor. We are confident that the doctor, once he has become aware that genetic problems are often very complex indeed, will preferably consult the specialist too often rather than too little. Even if the doctor has come to the conclusion that he cannot responsibly counsel any such case, the information in this guide will help him to understand the nature of the problem with which the specialist is confronted. It will, moreover, help him to recognize problems which require the specialist and allow him to aid the latter in gathering relevant information.

In the first chapter ("Appearances deceive") we have illustrated many of the problems of analysis with a specific case history. This approach necessitated the use of various terms and concepts, the exact meaning of which is defined and explained in later chapters. The reader is advised not to despair but to re-read the first chapter after

he has carefully read the rest. We recommend Chapter II as the starting point and Chapter I as the conclusion for those readers who have had little or no previous contact with genetics.

This guide is the result of long years of discussion in Berlin and more specifically of a close collaboration in Heidelberg, from January 1964 until August 1967. We sincerely hope that it will prove helpful to many practicing physicians and beneficial for patients and their families.

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