Molecular Diagnosis of Genetic Diseases
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Molecular Diagnosis of Genetic Diseases

Edited by
Rob Elles

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Preface

Many previous volumes concerned with methodology in human genetics have been written by research scientists and naturally reflect that culture. *Molecular Diagnosis of Genetic Diseases* aims to diverge from previous titles by presenting contributions that cover a key method in detail, but are set in the context of a diagnostic area or genetic disease. In this format, the book attempts to cover nearly all of the most common genetic disease diagnostics that are offered as services by clinical molecular genetics laboratories, thus contributing a reasonably comprehensive handbook for this type of center. Most of the authors are active scientists working in clinical diagnostics. The methods reflect their working experience in attempting to assure robust, reliable results, and to include essential controls, quality standards, and interpretive guides.

*Molecular Diagnosis of Genetic Diseases* is primarily aimed at scientists, clinicians, and technologists working in clinical molecular genetics, especially those working in, or with, diagnostic laboratories. Others who will find the book useful include students and scientific workers at the interface of research genetics and diagnostics, forensic scientists, and biotechnologists. Those concerned with the commercial development of the diagnostic field and with regulation or improvement in standards in molecular genetics, both in professional bodies or government agencies, will also be interested in this book. In addition, scientists planning to develop novel molecular genetic diagnostics in countries with little or no experience in this field will find the book a useful starting point.

The chapters form a practical guide to the introduction of new diagnostic areas into the laboratory and provide a bench book for day-to-day use and the development of laboratory-specific standard operating procedures. Finally, chapters have been included with the notion of stimulating fresh thinking about molecular genetic diagnostics in the context of risk analysis and genetic counseling and to consider the internal and external quality standards that will have to apply for the public and patients to have confidence in genetic testing.

Rob Elles
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