Book Review

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Guide to Mutation Detection. Edited by Graham R. Taylor and Ian N. Day. Hoboken, NJ: Wiley, 2005. Pp. 330. $89.95, paperback.

Guide to Mutation Detection is a compilation of short reviews and protocols for detecting changes in DNA sequence. Its intended audience is postgraduate-level molecular geneticists, particularly individuals working in diagnostic molecular-genetics laboratories. The book originates from a practical course that is offered once every 2 years in conjunction with the Human Genome Organization (HUGO) Mutation Detection Workshop. The reviews and protocols are contributed by the instructors of that course. The book is divided into two sections, Reviews and Methods. The Reviews section begins with general reviews and then progresses to more-specific technical reviews of methods for detecting DNA sequence changes. The Methods section offers more-detailed protocols for various methods, some of which are discussed in the Reviews section. In addition to wet-bench techniques, *Guide to Mutation Detection* has a chapter devoted to quality control and quality assurance in clinical laboratories. It also has several chapters dedicated to the use of both general mutation databases and locus-specific databases.

Guide to Mutation Detection covers a variety of topics that are of interest to clinical molecular-genetics laboratories. It is not a comprehensive review but covers selected techniques. The techniques range from the basic, such as PCR and Southern-blot analysis, to more cutting-edge methods, such as tag array minisequencing. The first few chapters in the Reviews section provide an excellent introduction to the nature of mutations encountered in clinical molecular-genetics laboratories. These chapters also describe some of the methods used to detect DNA mutations. More-detailed reviews of selected topics follow the two introductory chapters. The topics covered in the Reviews section are not a comprehensive list of methods but are only a subset of the methods used in diagnostic molecular-genetics laboratories. Surprisingly, there is little attention given to what is considered the gold standard for mutation detection, dye terminator DNA sequencing.

The Methods section provides detailed protocols for some of the topics discussed in the Reviews section as well as additional protocols for methods not covered there. Again, the topics range from the basic to more-novel methods. Like the Reviews section, the Methods section is not a comprehensive list of methodologies available. There is substantial variation in the quality and detail provided in these chapters. In fact, some of the chapters in the Methods section are essentially the protocols provided by the manufacturers. Other chapters, however, provide detailed protocols for techniques that have been developed after many years of experience by leaders in the field.

Guide to Mutation Detection is a relatively short manual, only 330 pages in length; hence, it is not a comprehensive review, as is *Current Protocols in Human Genetics*. The strongest feature of *Guide* is that it provides a good introduction to some cutting-edge technologies, such as the use of microarrays and tandem mass spectrometry in the analysis of DNA mutations. It also contains some protocols that have been developed by well-established laboratories that may be useful in detecting specific classes of mutations. For these reasons, *Guide to Mutation Detection* is a useful tool to complement more-comprehensive technical manuals.

BRADFORD COFFEE

Department of Human Genetics
Emory University School of Medicine
Atlanta

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