Preface

In the preface to the first edition of *PCR Mutation Detection Protocols*, we indicated that it was an exciting time for molecular genetics and, in particular, the use of molecular diagnostic techniques for the analysis of DNA and mutations. A number of years on and this has proven to be true and has been reflected in the numerous developments and methods that allow mutation analysis to be currently undertaken. The explosion of DNA sequence information, the access to bioinformatics and mutation databases coupled with the ability to readily detect and confirm mutations has cemented the role of molecular diagnostics in medicine. In particular, mutation detection by the PCR has stood the test of time and has been increasingly adopted by many laboratories and now forms the cornerstone of many DNA diagnostic techniques. Indeed, it is now very difficult to think of molecular diagnostics without the PCR, and it is interesting to see the many ways in which the PCR has been adapted and has evolved over the years. However, the methods that one would think of as having been replaced by the PCR are still very much with us and fulfill an important role. It is a testament to the developers of methods such as Southern blotting that they are part of the panel of techniques with which to identify mutations in DNA. It is, of course, important to include these techniques in this second edition of *PCR Mutation Detection Protocols* as well as the PCR and its many various incarnations such as SSCP, CSGE, and dHPLC.

One theme that is increasingly running through clinical diagnostic laboratories nowadays is demand for accurate diagnostics with high throughput. The increasing casework in many ways reflects the success of molecular diagnostic techniques and their adoption into the diagnostic armory of the clinician. With this in mind, the inclusion of a number of these methods in the second edition of *PCR Mutation Detection Protocols* is an important one. The increase in DNA analysis requests will no doubt lead to further developments especially in the PCR; however, it is the development of affordable, durable, and accurate microarray systems and their adoption into the clinical laboratory that will ultimately be able to cope with this demand, and a collection of such methods are presented in this edition.

As with the first edition of *PCR Mutation Detection Protocols*, each chapter includes the underlying basis of the method and allows the reader to select and undertake the method successfully. The notes sections with each of the chapters provides the often hard to find information that may mean the difference between success and failure of the method. *PCR Mutation Detection Protocols* is aimed at postgraduate scientists, researchers, and clinicians already engaged in the area. However, it may also provide an important first step for those wanting to adopt a new technique in their laboratory.

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