Preface

Alongside rapid advances in scientific understanding about cancer genomics, there have been huge steps forward in genetic testing for pathogenic mutations in cancer predisposing genes, as well as the management of cancer risks associated with these mutations.

Until recently, families with a history of cancer suggesting high risk cancer predisposition genes as their cause have been counselled and managed within specialised genetic services. As the number of individuals eligible for cancer predisposition testing is rapidly increasing and more management options and treatments tailored to pathways disrupted by mutated cancer predisposition genes are developed, oncologists, surgeons and other healthcare specialists treating these patients have to become more involved in genetic testing and managing cancer risks in their patients.

Much has been written about the diagnosis and management of patients with common hereditary cancer such as breast/ovarian and colorectal cancer syndromes. However, there is limited information available to health professionals who diagnose and manage rare hereditary cancer syndromes, some of which present in childhood.

This book approaches the issue of the differential diagnosis and management of rare hereditary cancer syndromes from a practical angle, addressing the issues for each tumour type as seen by health professionals in their day-to-day practice.

The first chapter aims to update cancer specialists on the newest developments in genetic testing technology. It describes the strengths, limitations and caveats of these technologies to enable cancer specialists to use these tests safely and effectively for the benefit of their patients.

The subsequent chapters describe how patients with specific rare hereditary cancer syndromes may be identified through their personal and family history of cancer, which genes should be tested based on these criteria, the clinical picture of the respective cancer syndromes caused by mutations in these genes, as well as the appropriate management options.

The final chapter deals with the wider issues involved in genetic counselling and testing for cancer susceptibility for patients, families and health professionals.

In summary, this book has been written by leading specialists in the field to enable health professionals to correctly identify patients with these rare syndromes who will benefit from genetic counselling and testing and to provide them with the knowledge to manage patients and advise family members who may be at risk of an inherited cancer predisposition.

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