

Molecular Diagnostics in Cytopathology

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Editors

Molecular Diagnostics in Cytopathology

A Practical Handbook for the
Practicing Pathologist



Springer

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Preface

This book is a compilation of high-yield molecular pathology topics relevant to the field of cytopathology in a user-friendly format that can be used as a reference handbook by training and practicing pathologists and laboratory personnel dealing with, and interested in, this evolving field.

Molecular diagnostics are increasingly used to help guide targeted therapy in solid organ tumors and hematologic malignancies. A large proportion of molecular testing is performed on limited-volume samples obtained via minimally invasive techniques, such as fine needle aspiration. Increasingly, cytopathologists play an essential role in this process, both in the triage of specimens during rapid on-site evaluation and in the evaluation of archival samples to determine suitability for ancillary testing. Therefore, it is imperative that practicing cytopathologists stay abreast of up-to-date diagnostic, prognostic, and predictive ancillary tests that can be used on limited cytologic material. This is a challenge since the landscape of known genomic alterations is constantly evolving and the subsequent set of testing options is ever expanding. In addition, many practicing cytopathologists have not had substantial molecular pathology training during residency or fellowship; therefore, the basic core principles of molecular testing may remain elusive.

The main focus of this book is to provide an overview of the principles of molecular diagnostics in context of cytopathology specimens together with a basic understanding and working knowledge of the available technology, platforms, and clinical applications. The initial sections of the book summarize the pre-analytic aspects of molecular testing, including

cytology specimen preparation and handling, specimen selection and evaluation, and workflow algorithms as well as the analytic considerations for a variety of nucleic acid and protein-based testing. The remaining section focuses on disease-specific molecular testing for various organ-based applications.

Molecular cytopathology is an expanding and evolving field. With the increasing demand for molecular testing on small specimens, cytopathologists need to understand how to efficiently select and triage the best tissue for testing as well as interpret the molecular test results in context of the morphology to cement the role of cytopathology as an independent and essential component of diagnostic and precision medicine.

We hope this book will serve as a practical handbook of clinical molecular diagnostics in context of the cytopathology specimen.

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