
Preface

Since the publication of the second edition of *Mitochondrial DNA: Methods and Protocols*, major technological advances have been made in high-throughput DNA sequencing, high-resolution cellular imaging, and the use of targeted nucleases for genetic manipulation. These new scientific tools have revolutionized the study of human genetics and biology and apply equally to the investigation of the mitochondrial genome. This has allowed researchers to gain new insights into how mitochondrial DNA modulates mitochondrial metabolism and also the association between mitochondrial DNA variation and human disease.

This third edition of *Mitochondrial DNA: Methods and Protocols* describes these new methodologies for mitochondrial DNA analysis and, combined with well-established protocols that are considered the gold standard in the field, comprises a compendium of detailed methods for the study of mitochondrial DNA biology. The first part describes protocols for detecting mutations in mitochondrial DNA by next-generation massive parallel sequencing as well as techniques to assess mitochondrial DNA damage. The second part describes the visualization of mitochondrial DNA in situ and the detection of mitochondrial DNA nucleoids within the mitochondria. The third part describes methods for analyzing mitochondrial DNA replication, mitochondrial DNA-encoded protein translation, and mitochondrial DNA copy number. The fourth part describes the latest technology for modifying the mitochondrial genome, while the fifth part describes how to purify proteins involved in the replication and transcription of mitochondrial DNA.

These protocols will prove highly useful, not only for mitochondrial researchers but also for scientists studying human diseases where mitochondrial DNA variation has been recognized as an important pathogenic factor, including cancer and neurodegeneration. As such, this book will be a valuable addition to the *Mitochondrial DNA: Methods and Protocols* series title, and I would like to thank all of the authors for their excellent contributions to this publication.

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<http://www.springer.com/978-1-4939-3039-5>

Mitochondrial DNA

Methods and Protocols

McKenzie, M. (Ed.)

2016, XI, 234 p., Hardcover

ISBN: 978-1-4939-3039-5

A product of Humana Press