

Preface

The transmission, segregation, and inheritance of mitochondrial DNA (mtDNA) have been increasingly investigated over the last 20 years. mtDNA is transmitted through the oocyte, embryo, and fetus to the live offspring. However, before this mtDNA can be transmitted, the whole process is initially dependent on the quality of the oocyte prior to fertilization and the fertilizing sperm. The first section of this volume involves extensive discussion of the importance of sperm and oocyte mitochondria in the processes prior to fertilization and elicits their contributions to the fertilization process. It then discusses the role of mitochondria during preimplantation development and how mtDNA contributes to this process. It also defines how transcription and replication of this genome are tightly regulated during preimplantation embryogenesis.

The second section concentrates on mtDNA transmission through the oocyte. It draws on findings from animal and cellular experimental model systems to explain how mutated mtDNA are segregated and can consequently result in severely debilitating disease or can even be fatal. The rationale for such scientific investigation is supported by a detailed account documenting and discussing the impact, etiology, and pathology of human mitochondrial disease.

Section three describes in detail the several assisted reproductive techniques that have been proposed to prevent the transmission of mitochondrial disease. It also demonstrates how these techniques can be successfully tested and developed prior to potential consideration in the clinical environment. It is followed by a discussion of various screening approaches that might be utilized to determine whether a woman is at risk of transmitting a mitochondrial disease to her offspring prior to and shortly after implantation.

The final section concentrates on the problems associated with some of these sophisticated and controversial reproductive technologies. It discusses the potential that these techniques have for violating the strict unimaternal transmission of mtDNA and its highly regulated transcription and replication mechanisms. It also raises concerns about the use of such techniques for the generation of tailor-made embryonic stem cells for patient-specific cell therapies and suggests mechanism at the mitochondrial level for overcoming such anomalies.

The investigators contributing to each of these sections are experienced and accomplished. They have provided a wealth of knowledge to provide an all encompassing account of mtDNA from the oocyte to the live offspring.

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