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Exceptional Child Education Resources ,1998

Mitochondrial Disease Genes Compendium Marni J. Falk, M.D.,2020-05-13 The field of Mitochondrial Medicine has been dominated by symptom constellation-based diagnostic categorization since the first clinical syndrome was described three decades ago. Now, as rapidly expanding knowledge has revealed that mitochondrial diseases may result from several hundred distinct gene disorders with extensive clinical and mutation heterogeneity, the most useful guide for clinical care and research embraces a gene-centric approach to each individual's disorder. Together with international colleagues, Dr. Marni Falk has developed the Mitochondrial Disease Sequence Data Resource (MSeqDR), an online, community curated, centralized data resource of mitochondrial disease data from a genomic perspective. MSeqDR provides tens of thousands of users with interactive access to mitochondrial disease feature descriptions in a defined human phenotype ontology, mitochondrial proteome-based gene and variant curation, and a suite of easily accessible tools to facilitate analysis of complex genomic datasets in nuclear and mitochondrial genomes as well as accurate interpretation of mitochondrial disease genes, variants, and diseases. Here, in the Mitochondrial Disease Genes Compendium, Dr. Marni Falk and a team of international experts have built off their work on MSeqDR to provide an all-in-one, readily accessible, and easy-to-use at point of care reference on mitochondrial disease from a gene-based perspective. In this book, clinicians and researchers will find a complete overview of mitochondrial disease genes relevant across all specialties, cataloging and building context around clinical features and the genetic basis of each condition. Within, each gene page offers an in-depth, referenced view of the relevant clinical disease spectrum, including gene and protein descriptions, year discovered, inheritance pattern(s), age ranges affected, major clinical features and severity range, clinical pearls, known therapies, available support groups, animal models, and gene-specific basic, translational, or clinical research activities now underway. Links provided on each gene page direct readers to MSeqDR for new findings, up-to-date genomic variant data, and user friendly informatics tools accessible to general clinicians and sophisticated geneticists or bioinformaticians alike, ensuring access to updated information on each condition. Covering 256 mitochondrial disease genes that have been expert-curated to assure they cause human diseases and have known mitochondrial localization or impact mitochondrial function, the Mitochondrial Disease Genes Compendium is directed at clinicians and researchers, facilitating bedside access to high-level, curated knowledge on mitochondrial disease genetics that rapidly enables patient diagnosis, counseling, management, treatment, and research. Provides a readily intelligible, all-in-one reference of known mitochondrial disease genes and their associated conditions Features live links to Mitochondrial Disease Sequence Data Resource (MSeqDR) pages with regularly updated genetic variant data and bioinformatics tools Covers the inheritance patterns, age spectrum affected, major clinical features, therapeutics, support groups, and research currently under way for over 250 mitochondrial diseases

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