Mitochondrial disease

Defects of mitochondrial metabolism cause a wide range of human diseases that include examples from all medical subspecialties. This review updates the topic of mitochondrial diseases by reviewing the most important recent advances in this area. The factors influencing inheritance, maintenance and replication of mtDNA are reviewed and the genotype-phenotype of mtDNA disorders has been expanded, with new insights into epidemiology, pathogenesis and its role in ageing. Recently identified nuclear gene mutations of mitochondrial proteins include mutations of *frataxin* causing Friedreich's ataxia, *PINK1, DJ1* causing Parkinson's disease and *POLG* causing infantile mtDNA depletion syndrome, ophthalmoplegia, parkinsonism, male subfertility and, in a transgenic mouse model, premature senescence. Mitochondrial defects in neurodegenerative diseases include Parkinson's, Alzheimer's and Huntington's disease. Improved understanding of mtDNA inheritance and mutation penetrance patterns, and novel techniques for mtDNA modification offer significant prospects for more accurate genetic counselling and effective future therapies.

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