Title (en)

HUMAN MITOCHONDRIAL DNA POLYMORPHISMS, HAPLOGROUPS, ASSOCIATIONS WITH PHYSIOLOGICAL CONDITIONS, AND GENOTYPING ARRAYS

Title (de)

MENSCHLICHE MITOCHONDRIALE DNA-POLYMORPHISMEN, HAPLOGRUPPEN, ASSOZIATIONEN MIT PHYSIOLOGISCHEN LEIDEN SOWIE GENOTYPISCHE ARRAYS

Title (fr)

POLYMORPHISMES D'ADN MITOCHONDRIAL HUMAIN, HAPLOGROUPES, ASSOCIATIONS AVEC DES CONDITIONS PHYSIOLOGIQUES ET RESEAUX DE GENOTYPAGE

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Abstract (en)

[origin: WO03018775A2] This inention provides human mtDNA polymorphisms that are diagnostic of all the major human haplogroups and methods of diagnosing those haplogroups and selected subhaplogroups. This invention also provides methods for identifying evolutionarily significant mitochondrial DNA genes, nucleotide alleles, and amino acid alleles. Evolutionarily significant genes and alleles are identified using one or two populations of a single species. The process of identifying evolutionarily significant nucleotide alleles involves identifying evolutionarily significant genes and then evolutionarily significant nucleotide alleles in those genes, and identifying evolutionarily significant amino acid alleles involves identifying amino acids encoded by all nonsynonymous alleles. Synonymous codings of the nucleotide alleles encoding evolutionarily significant amino acid alleles of this invention are equivalent to the evolutionarily significant amino acid alleles disclosed herein and are included within the scope of this invention. Synonymous codings include alleles at neighboring nucleotide loci that are within the same codon. This invention also provides methods for associating haplogroups and evolutionarily significant nucleotide and amino acid alleles with predispositions to physiological conditions. Methods for diagnosing predisposition to LHON, and methods for diagnosing increased likelihood of developing blindness, centenaria, and increased longevity that are not dependent on the geographical location of the individual being diagnosed are provided herein. Diagnosis of an individual with a predisposition to an energy metabolism-related physiological condition is dependent on the geographic region of the individual. Physiological conditions diagnosable by the methods of this invention include healthy conditions and pathological conditions. Physiological conditions that are associated with haplogroups and with alleles provided by this invention include energetic imbalance, metabolic disease, abnormal energy metabolism, abnormal temperature regulation, abnormal oxidative phosphorylation, abnormal electron transport, obesity, amount of body fat, diabetes, hypertension, and cardiovascular disease.

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Citation (search report)

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- See references of WO 03018775A2

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