Multiplex analysis of mitochondrial DNA pathogenic and polymorphic sequence variants

Titre	Multiplex analysis of mitochondrial DNA pathogenic and polymorphic sequence variants
Type de publication	Article de revue
Auteur	Poole, J. C [1], Procaccio, Vincent [2], Brandon, M. C [3], Merrick, G. [4], Wallace, D. C [5]
Editeur	De Gruyter
Type	Article scientifique dans une revue à comité de lecture
Année	2010
Langue	Anglais
Date	2010
Numéro	10
Pagination	1115 - 30
Volume	391
Titre de la revue	Biological Chemistry
ISSN	1431-6730
Mots-clés	Alleles [6], Cell Line, Tumor [7], DNA Primers/genetics [8], DNA, Mitochondrial/genetics [9], Haplotypes [10], Humans [11], internet [12], Phylogeny [13], Polymerase Chain Reaction/methods [14], Polymorphism, Single Nucleotide/genetics [15], Pseudogenes/genetics [16]
Résumé en anglais	The mitochondrial DNA (mtDNA) encompasses two classes of functionally important sequence variants: recent pathogenic mutations and ancient adaptive polymorphisms. To rapidly and cheaply evaluate both classes of single nucleotide variants (SNVs), we have developed an integrated system in which mtDNA SNVs are analyzed by multiplex primer extension using the SNaPshot system. A multiplex PCR amplification strategy was used to amplify the entire mtDNA, a computer program identifies optimal extension primers, and a complete global haplotyping system is also proposed. This system genotypes SNVs on multiplexed mtDNA PCR products or directly from enriched mtDNA samples and can quantify heteroplasmic variants down to 0.8% using a standard curve. With this system, we have developed assays for testing the common pathogenic mutations in four multiplex panels: two genotype the 13 most common pathogenic mtDNA mutations and two genotype the 10 most common Leber Hereditary Optic Neuropathy mutations along with haplogroups J and T. We use a hierarchal system of 140 SNVs to delineate the major global mtDNA haplogroups based on a global phylogenetic tree of coding region polymorphisms. This system should permit rapid and inexpensive genotyping of pathogenic and lineage-specific mtDNA SNVs by clinical and research laboratories.
URL de la notice	http://okina.univ-angers.fr/publications/ua8337 [17]
DOI	10.1515/BC.2010.125 [18]

Lien vers le document	http://dx.doi.org/10.1515/BC.2010.125 [18]
Titre abrégé	Biol Chem

Liens
[1] http://okina.univ-angers.fr/publications?f[author]=14408
[2] http://okina.univ-angers.fr/v.procaccio/publications
[3] http://okina.univ-angers.fr/publications?f[author]=14409
[4] http://okina.univ-angers.fr/publications?f[author]=14410
[5] http://okina.univ-angers.fr/publications?f[author]=14289
[6] http://okina.univ-angers.fr/publications?f[keyword]=11219
[7] http://okina.univ-angers.fr/publications?f[keyword]=1099
[8] http://okina.univ-angers.fr/publications?f[keyword]=12972
[9] http://okina.univ-angers.fr/publications?f[keyword]=12971
[10] http://okina.univ-angers.fr/publications?f[keyword]=13635
[11] http://okina.univ-angers.fr/publications?f[keyword]=991
[12] http://okina.univ-angers.fr/publications?f[keyword]=2631
[13] http://okina.univ-angers.fr/publications?f[keyword]=8736
[14] http://okina.univ-angers.fr/publications?f[keyword]=13636
[15] http://okina.univ-angers.fr/publications?f[keyword]=12866
[16] http://okina.univ-angers.fr/publications?f[keyword]=13637
[17] http://okina.univ-angers.fr/publications/ua8337
[18] http://dx.doi.org/10.1515/BC.2010.125