J Med Genet. 2012 Apr;49(4):234-41.

Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes.

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Abstract

OBJECTIVE: To investigate the utility of autozygome analysis and exome sequencing in a cohort of patients with suspected or confirmed mitochondrial encephalomyopathy.

METHODS: Autozygome was used to highlight candidate genes for direct sequencing in 10 probands, all born to consanguineous parents. Autozygome was also used to filter the variants from exome sequencing of four probands.

RESULTS: In addition to revealing mutations in known mitochondrial genes, the analysis revealed the identification of two novel candidate disease genes: MFF and FARS2, encoding the mitochondrial fission factor and phenylalanyl-tRNA synthetase, respectively.

INTERPRETATION: These findings expand the repertoire of genes that are mutated in patients with mitochondrial disorders and highlight the value of integrating genomic approaches in the evaluation of these patients.