

UKPDS 21: low prevalence of the mitochondrial transfer RNA gene (tRNA(Leu(UUR))) mutation at position 3243bp in UK Caucasian type 2 diabetic patients.

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Some patients with Type 2 (non-insulin-dependent) diabetes mellitus possess a mitochondrial mutation in the tRNA(Leu(UUR)) gene at position 3243 bp. These subjects show a maternal mode of inheritance and often have hearing defects. In French and Japanese populations, this mutation may be present in 1-3% of subjects with a family history of diabetes. We assessed the prevalence of this mutation in newly diagnosed diabetic subjects in the UK white Caucasian population. The 3243 bp mutation was not detected in 500 randomly selected Type 2 diabetic subjects, 50 gestational diabetic subjects, and members of a MODY pedigree. Two of 748 (0.27%) Type 2 diabetic subjects with a family history of diabetes were found to possess the mutation. These subjects had an early age of diagnosis (M 38 years; F 36 years) and were non-obese. The male patient showed evidence of markedly impaired beta-cell function and deafness, while the female was not deaf, had approximately 50% of normal pancreatic function and responded well to diet. The mutation in the tRNA(Leu(UUR)) gene probably occurs in only approximately 0.1-0.2% of white Caucasian Type 2 diabetic patients in the UK.