

## Mitochondrial DNA A3243G mutation involved in familial diabetes, chronic intestinal pseudo-obstruction and recurrent pancreatitis

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R�sum� en anglais	<p>Aims To report on a family with five members who carry the A3243G mutation in mitochondrial tRNA for leucine 1 (MTTL1) and present with diabetes, chronic intestinal pseudo-obstruction (CIPO) and recurrent pancreatitis, and to screen for this mutation in a cohort of 36 unrelated patients with recurrent pancreatitis.</p> <p>Methods The mutation was quantified in several tissue samples from patients. Respiratory chain activity was studied in muscle biopsies and fibroblast cultures. In addition, the thymidine phosphorylase gene (TP) involved in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) and three genes involved in chronic pancreatitis - PRSS1, SPINK1 and CFTR - were sequenced in affected patients. Finally, the MTTL1 gene was examined in 36 unrelated patients who had recurrent pancreatitis, but no mutations in the PRSS1 and SPINK1 genes. Results Heteroplasmy for the mtDNA A3243G mutation was found in all tissue samples from these patients, but no mutations were found in the genes coding for thymidine phosphorylase, PRSS1, SPINK1 and CFTR. Also, none of the 36 unrelated patients with recurrent pancreatitis were carrying any MTTL1 mutations. Conclusion The mtDNA A3243G mutation associated with the gastrointestinal manifestations observed in the affected family should be regarded as a possible cause of CIPO and unexplained recurrent pancreatitis. However, the mutation is probably only weakly involved in cases of isolated recurrent pancreatitis.</p>
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