

ASSOCIATIONS OF MITOCHONDRIAL HAPLOGROUPS WITH BILIARY ATRESIA

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Background:

Various lines of evidence demonstrate the involvement of mitochondrial dysfunction in the pathogenesis of biliary atresia (BA). However, the precise role of mitochondria in the liver cascade leading to BA is still unclear. Mitochondrial DNA (mtDNA) haplogroups-specific polymorphisms were not previously studied the association with BA.

Aim:

This study was to determine whether mtDNA polymorphism(s) play the role of "modifier gene(s)" in BA.

Methods:

In this work we have performed haplogroup analysis. 67 mitochondrial single nucleotide polymorphisms of 31 major mitochondrial haplogroups were determined by use of 24-plex PCR and fluorescent beads combined with sequence-specific oligonucleotide probes. The genotypes were predefined Japan mtDNA haplogroups in 77 BA patients and 404 controls.

Results:

Significantly higher prevalence of haplogroup E was detected in BA than in the controls (OR 4.764 95% CI 1.25-18.20, $p=0.032$). Lower prevalence of haplogroups B4 was detected in BA than in the controls (OR 0.84 95% CI 0.81-0.88, $p=0.014$).

Conclusions:

Our study support that the association between mtDNA haplogroup E and BA.

Key words:

mitochondria, haplogroup, BA