G73 A Preliminary and Pilot Study About Mitochondrial DNA Deletion in Sudden Infant Death Syndrome: An Endemic Study in Taiwan

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After attending this presentation, attendees will gain an understanding whether there is a relationship between mitochondrial DNA deletion and sudden infant death syndrome. This presentation will impact the forensic community and/or humanity by demonstrating how although some change of genetic markers in mitochondrial DNA may not be the real etiological cause of death in SIDS cases; it could be a contributing factor to an infant's death within a critical medical condition or situation.

Sudden infant death syndrome (SIDS) is a leading cause of postneonatal infant mortality and a serious and challenging issue confronting the medical and legal professions. Many hypotheses have been proposed and studied, however, a consensus on the causes of SIDS is still lacking. Although a number of coding region mitochondrial DNA (mtDNA) mutations involving SIDS have been reported, the role of mtDNA deletion or depletion in SIDS victims is still unknown. This study was designed to investigate whether mtDNA deletions exert any effect on the etiology of SIDS. Statistical data have shown that infants dying from SIDS score lower in activity tests and appear to be sleepier and less reactive than control subjects. These behavioral characteristics may have been the result of ATP depletion attributable to mutations or deletion in mtDNA.

Seven SIDS and 19 non-SIDS fatalities were included in this study to determine the relative amount of mtDNA copy number and the occurrence of mtDNA deletion in blood, skeletal muscle, and cardiac muscle specimens. Analytical approaches included real-time quantitative PCR, primer-shift PCR analysis and DNA sequencing. Breakpoints of the three types of mtDNA deletions (4977, 5335, and 7599 bp deletions) observed in the population were identified by sequencing methods.

Only one specimen (cardiac muscle) from a congenital heart malformation subject was found to have 4977 bp mtDNA deletion. Fisher's exact probability test and Spearman's correlation coefficient were applied to the analysis of the observed data on 5335 bp and 7599 bp mtDNA deletions and found no statistically significant difference on the occurrence frequencies of 5335 bp and 7599 bp mtDNA deletions between SIDS and nonSIDS victims. However, the observed data indicate: (a) for blood specimens, the occurrence frequencies of 5335 and 7599 bp mtDNA deletion observed in the SIDS were 4and 2-fold, respectively, higher than the nonSIDS victims; (b) for skeletal muscle specimens, the occurrence frequencies of 5335 bp mtDNA deletion in SIDS victims was 1.8-fold of the non-SIDS victims. No significant correlation was observed on the relative amount of mtDNA copy number and the occurrence frequencies of mtDNA deletions between the SIDS and non-SIDS groups; however, the occurrence frequencies of 5335 bp and 7599 bp deletions and the relative amount of mtDNA copy number in the skeletal and cardiac muscle specimens from the SIDS group were much higher than that from the non-SIDS group.

The increase in mtDNA content in mtDNA deletion cases correlates with mitochondrial proliferation that might have been a compensatory mechanism of defective mitochondria. These defects in mtDNA may result in impaired production of ATP and bioenergetic crisis. mtDNA deletions in themselves do not cause SIDS but may cause energy deficiency or hypoxia in stressful situation during a vulnerable developmental stage. These preliminary results show that mitochondrial DNA deletion might predispose an infant to death in a critical medical situation.

Sudden Infant Death Syndrome, SIDS, Mitochondrial DNA Deletion

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