Newborn Screening, Inborn Errors of Metabolism

AB062. DNA damage is associated with infantile-pediatric cases of mitochondrial disorders: a pilot study from North India

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Background: High oxidative stress increases the pathogenicity of mitochondrial disorders by increases the production of ROS/free radicals, results in compromising DNA (mitochondrial & nuclear DNA) stability at the basal level. Aim of this study was to decipher the percentage of DNA damage in different age groups of patients with mitochondrial disorders.

Methods: Human leukocytes isolated from whole blood samples of 35 patients, including 10 cases of fatal infantile lactic acidosis (FILA), 10 cases of early pediatric age groups (4 mitochondrial myopathy, 3 MELAS, 2 Leigh disease & 1 PEO), and 15 from late pediatric age group. Samples from 20 healthy controls were also included. Each sample was mixed in phosphate buffered saline (PBS) and processed for the alkaline comet assay. More than 500 nuclei in controls & cases, scanned using fluorescence microscope (Metafer 4), calculated automatically via Meta-Cyte Comet Scan system for tail length, tail movement, olive tail movement and percentage DNA in tail.

Results: All specimens from the FILA and pediatric early age groups showed very high percentage of DNA damage as compared to healthy control samples.

Conclusions: This pilot study revealed significantly high percentage of DNA damage among samples from infantile and early pediatric subpopulation study compared to healthy control samples, thus significantly correlated with the disease severity of mitochondrial disorders in infantile-pediatrics population. Screening for DNA damage in mitochondriopathy at early age groups can helps us to amend the natural course of disease by minimizing the morbidity & mortality through better preventive medication and therapy.

Keywords: Fatal infantile lactic acidosis (FILA); MELAS; ROS

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