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Analysis of Epigenetic Modification in Leber's Hereditary Optic Neuropathy (LHON) Cells

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ABSTRACT Leber's Hereditary Optic Neuropathy (LHON) is a mitochondrially inherited vision loss disease due to pathogenic mitochondrial mutations in complex 1 encoding genes which are incurable to date. Mitochondrial respiratory chain complex 1 consists of 45 subunits and *NDUFS4* is a nuclear-encoded accessory submit which has a significant role in mitochondrial complex 1 assembly. This preliminary study focuses on the histone modification changes in the *NDUFS4* gene. For this study chromatin immunoprecipitation (ChIP) assay was performed using LHON *ND4* mutant cell lines and PBMC from a healthy control. Five histone modification antibodies such as $H_3K_{18}Ac$, $H_3K_{27}Ac$, $H_3K_9Me_2$, $H_3K_4Me_3$, and $H_3K_{27}Me_3$ were used for the experiment. ChIP-qPCR was performed to determine the histone enrichment in the *NDUFS4* promoter region. ChIP-qPCR data showed that H3K18Ac histone enrichment has variation in LHON *ND4* mutant cells compared to PBMCs. As a primary step, this study has tried to figure out the histone modification changes in the *NDUFS4* gene in the LHON cells derived from *ND4* mutant patients and control PBMCs. Epigenetic studies in nuclear-encoded mitochondrial proteins in LHON may help researchers for a better understanding of disease pathology.