Background: Acute lymphoblastic leukemia (ALL) is the most common cancer diagnosed in children and represents approximately 25% of cancer diagnoses among those younger than 15 years of age. Materials and Methods: This study investigated alterations in the displacement loop (d-loop) region of mitochondrial DNA (mtDNA) as a risk factor and diagnostic biomarker for early detection and diagnosis of acute lymphoblastic leukemia. Using mtDNA from 23 subjects diagnosed with acute lymphoblastic leukemia, the first 450 bp of the d-loop region were amplified and successfully sequenced. Results: This revealed 132 mutations at 25 positions in this region, with a mean of 6 alterations per subject. The d-loop alterations in mtDNA in subjects were all identified as single nucleotide polymorphisms in a homoplasmic distribution pattern. Mutant alleles were observed in all subjects with individual frequency rates of up to 95%. Thirteen mutant alleles in the d-loop region of mtDNA occurred with a high frequency. Novel alleles and locations were also identified in the d-loop of mtDNA as follows: 89 G insertions (40%), 95 G insertions (13%), 182 C/T substitutions (5%), 308 C insertions (19%), and 311 C insertions (80%). The findings of this study need to be replicated to be confirmed. Conclusions: Further investigation of the relationship between mutations in mitochondrial d-loop genes and incidence of acute lymphoblastic leukemia is recommended.

**Title:** Novel Mutations in the Displacement Loop of Mitochondrial DNA are Associated with Acute Lymphoblastic Leukemia: A Genetic Sequencing Study

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**Abstract:**

The focus of this study was to investigate the prevalence of mutations in the d-loop region of mitochondrial DNA (mtDNA) in patients with acute lymphoblastic leukemia (ALL). The study sample consisted of 23 subjects diagnosed with acute lymphoblastic leukemia. The first 450 bp of the d-loop region were amplified and successfully sequenced. This revealed 132 mutations at 25 positions in this region, with a mean of 6 alterations per subject. The d-loop alterations in mtDNA in subjects were all identified as single nucleotide polymorphisms in a homoplasmic distribution pattern. Mutant alleles were observed in all subjects with individual frequency rates of up to 95%. Thirteen mutant alleles in the d-loop region of mtDNA occurred with a high frequency. Novel alleles and locations were also identified in the d-loop of mtDNA as follows: 89 G insertions (40%), 95 G insertions (13%), 182 C/T substitutions (5%), 308 C insertions (19%), and 311 C insertions (80%). The findings of this study need to be replicated to be confirmed. Conclusions: Further investigation of the relationship between mutations in mitochondrial d-loop genes and incidence of acute lymphoblastic leukemia is recommended.