



# Insights of Somatic Mutations and their Implications in Lymphoblastoid Cell Lines

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## DESCRIPTION

Somatic mutations are changes in the Deoxyribose Nucleic Acid (DNA) of a cell that occur after conception and can be passed on to many copies of the original cell. Such mutations can cause diseases like cancer if they occur in important genes.

In lymphoblastoid cell lines, somatic mutations are important because they can affect the behavior and characteristics of these cells. Lymphoblastoid Cell Lines (LCLs) are established from human lymphocytes and used to study genetic variation, gene regulation, and cancer biology. Somatic mutations play a key role in the development and functioning of LCLs. It has been found that most of these somatic mutations occur in non-coding regions such as enhancers, silencers, promoters, and intergenic regions which influence gene expression by regulating transcription factor binding.

When somatic mutations take place in LCLs they can lead to genetic alterations which may result in altered phenotypes such as increased or decreased growth rate, changes in morphology or cell surface properties. Furthermore, somatic mutations have also been shown to affect gene expression levels as well as epigenetic modifications such as DNA methylation. In addition to affecting gene expression levels, somatic mutations may also lead to chromosomal rearrangements such as deletions or translocations which may alter gene expression profiles. The most common way to analyze the role of somatic mutations is through Whole Genome Sequencing (WGS). WGS has become an increasingly popular tool for analyzing genomic data due to its ability to detect Single Nucleotide Polymorphisms (SNPs), small insertions/deletions (indels), Copy Number Variants (CNVs), Structural Variants (SVs) and large rearrangements.

Somatic mutations play an important role in the development and functioning of lymphoblastoid cell lines. Through whole genome sequencing are able to better understand the impact of these mutations on gene expression levels and other cellular processes which could eventually lead to new treatments for various diseases. Somatic mutations refer to changes in genetic

material that occur in certain non-reproductive cells that are not passed on to offspring. The most common type of somatic mutation is a point mutation, which is a substitution, deletion or insertion of one or more nucleotides from a gene's DNA sequence. Other types of somatic mutations include chromosomal alterations such as deletions, duplications, and translocations. Chromosomal alterations happen when there is a change in the structure or number of chromosomes within a cell. These changes can be caused by errors during cell division or exposure to certain environmental agents. In lymphoblastoid cell lines, it has been observed that there can be hundreds of different kinds of somatic mutations. Somatic mutations are changes to a cell's genetic material that are not passed onto offspring or other cells.

They can result from exposure to environmental toxins, or even occur naturally through errors in DNA replication. These mutations can potentially alter how proteins interact with each other and disrupt the normal functioning of the cell. The study of somatic mutations has been crucial for understanding how cancer develops and progresses. Scientists have been able to use somatic mutation data from lymphoblastoid cell lines to uncover key pathways involved in tumor genesis, as well as predict patient outcomes. This information has been incredibly valuable for developing therapeutic strategies targeting specific molecular events that occur within a tumor. By examining the changes in gene expression patterns due to genetic alterations caused by somatic mutations can gain insight into age-related diseases like Alzheimer's and Parkinson's.

Overall, studying the impact of somatic mutations on cellular function in lymphoblastoid cell lines is an important area of yielding valuable information about many aspects of biology and disease pathology. It is clear that this will continue to be essential for advancing our understanding of human health. The role played by somatic mutations in lymphoblastoid cell lines, we are able to better understand how these cells develop and evolve over time. This knowledge can then be used to inform therapeutic strategies for treating cancerous tumors and other diseases that are caused by defective somatic mutations.

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