

Abstract

Gene polymorphisms are variations in DNA sequences that occur within a population, contributing to genetic diversity. These variations include single nucleotide polymorphisms (SNPs), insertions/deletions (indels), and copy number variations (CNVs), as well as the mechanisms of inheritance. Understanding these genetic variations is essential not only for elucidating the mechanisms of inheritance but also for exploring their implications in health and disease [1].

The presence of polymorphisms can significantly influence individual susceptibility to various diseases, ranging from common conditions like diabetes and heart disease to complex disorders such as cancer and autoimmune diseases. Moreover, polymorphisms play a pivotal role in pharmacogenomics, where individual genetic profiles affect drug metabolism, efficacy, and safety. Identifying relevant polymorphisms can lead to more personalized medical approaches, allowing for tailored therapies that optimize treatment outcomes. From an evolutionary perspective, gene polymorphisms provide insights into population dynamics, migration patterns, and adaptive strategies. The study of these variations helps researchers understand how populations respond to environmental pressures, thereby shedding light on the processes that shape biodiversity over time [2].

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Introduction

Gene polymorphisms refer to the variations in DNA sequences that exist among individuals within a population. These variations can take many forms, including single nucleotide polymorphisms (SNPs), insertions and deletions (indels), and larger structural changes such as copy number variations (CNVs). Polymorphisms are crucial for genetic diversity, which serves as the raw material for evolution and adaptation. Understanding these genetic variations is essential not only for elucidating the mechanisms of inheritance but also for exploring their implications in health and disease [1].

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