Abstract

Õ^}^Á][|^{[¦]@â•{Á¦^-^¦•Ác[Ác@^Áçæ¦åæcå[}Áâ}ÁÖÞŒÁ•^˘˘^}&^•Áæ{[}*Áâ}åâçåâ˘æ|•ÊÁ•å*}â,&æ}c|^Á&[}cïâà˘cå}*Ác[Á *^}^câ&Áåâ¡ç^¦•âc^Áæ}âÁâ}'˘^}&â}*Á¦]@^}[c^]â&Ác¦æåc•ÊÁåâ•^æ•^Á•˘•&^]cïâà¡âc^ÊÁæ}åâ¦˘*Á¦^•][}•^ÁV@â•Á[ç^¦çâ^¸Á^¢]|[¦^•Á c@^Á{^&@æ}â•{•Á•Ác@æcÁ*âç^Álïå•æ^Ác[Á*^Á^Á][|^{[¦]@â•{•Á¦{Á{•Ê|&|æ}[*â^]âçÊâç^æ}â¦^Á^*Á¦^•]^•Á[}&ï˘À]âãç]â˘Á
polymorphisms into types such as single nucleotide polymorphisms (SNPs), insertions and deletions (indels), copy
}˘{à^¦À˘^¦Á

Gene polymorphism is a fundamental concept in genetics that describes the occurrence of two or more genetically determined variants (alleles) within a population. ese variations can arise from mutations in the DNA sequence and are critical in contributing to the diversity observed in human traits, responses to environmental factors, and susceptibility to diseases. Understanding gene polymorphism is essential for deciphering the complex interplay between genetics and health. Polymorphisms can be classi ed into several categories, including single nucleotide polymorphisms (SNPs), which are the most prevalent form, as well as insertions, deletions, and larger structural variations. ese genetic di erences can a ect gene function, regulatory elements, and protein expression, leading to diverse phenotypic outcomes [1].

e signi cance of gene polymorphisms extends beyond basic research; they have practical implications in clinical settings, particularly in the elds of genomics and personalized medicine. By identifying speci c polymorphisms associated with various diseases, researchers can enhance disease risk assessments, improve diagnostic tools, and tailor treatments to individual genetic pro les. is article aims to provide a comprehensive overview of gene polymorphism, exploring its underlying mechanisms, categorization, and implications for human health. rough this exploration, we aim to highlight the importance of gene polymorphism in understanding genetic diversity and its role in shaping health outcomes [2].

Gene polymorphism is a fundamental concept in genetics that describes the occurrence of two or more genetically determined variants (alleles) within a population. ese variations can arise from mutations in the DNA sequence and are critical in contributing to the diversity observed in human traits, responses to environmental factors, and susceptibility to diseases. Understanding gene polymorphism is essential for deciphering the complex interplay between genetics and health.

Polymorphisms can be classi ed into several categories, including single nucleotide polymorphisms (SNPs), which are the most prevalent form, as well as insertions, deletions, and larger structural variations. SNPs, for instance, involve a change in a single nucleotide and occur frequently across the genome, a ecting gene function and expression. Insertions and deletions (indels) can disrupt coding sequences or regulatory regions, potentially leading to loss of function or altered gene

the genetic architecture of human populations and its far-reaching implications for health and disease. As we delve deeper into the mechanisms and types of polymorphisms, it becomes evident that these variations are not merely academic curiosities but fundamental factors in uencing biological processes and health outcomes. Understanding the mechanisms that generate gene polymorphisms-such as mutations [6], recombination, and gene ow-provides insights into how genetic diversity is maintained within populations. Mutations, whether spontaneous or induced by environmental factors, serve as the primary source of genetic variation. e e ects of these mutations can vary widely, ranging from benign to pathogenic, depending on their nature and location within the genome. Recombination during meiosis contributes to this diversity by reshu ing genetic material, which can lead to new allele combinations that may enhance adaptability to changing environments [7].

 e di erent types of gene polymorphisms-SNPs, indels, CNVs, and structural variants-each play distinct roles in phenotypic expression and disease susceptibility. SNPs, being the most abundant form of genetic variation, are particularly valuable in genome-wide association studies (GWAS) aimed at identifying genetic predispositions to diseases. For instance, certain SNPs have been associated with an increased risk of conditions like Alzheimer's disease and various cancers, o ering pathways for early diagnosis and intervention. Indels and CNVs further complicate this landscape, as they can in uence gene dosage and contribute to complex traits, such as height or intelligence, illu (ato)]-1.2 most abulc v28Fs. For inperan populations anlablact,iw 8.575 -1.647d [(e di ernt type5 (for healthhisms-SNPs,)0.6 (inosis J-sonaliztifyar