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Short Communication

Genetic Variations & It's Impact

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ABSTRACT

An organism's gene pool has a diverse variety of genetic variants. Variations have been linked to illnesses such as cancer, Turner syndrome, sickle cell anaemia, cystic fibrosis, among others, and some of them have also been shown to be helpful in some circumstances, such as raising bone density, reducing cholesterol, and improving malaria resistance. The significance of genetic changes or switches in human development has also been investigated. Monogenic diseases are genetic illnesses caused by single-gene mutations.

KEYWORDS: Down syndrome, Trisomy 21, Chromosome abnormality.

INTRODUCTION

A single nucleotide is a single strand of DNA. Some types of genetic mutations include polymorphism, structural variations, and genomic rearrangements. Rare diseases are caused by defects in a gene's DNA pattern, which is one of the reasons blamed for their occurrence. Although mutation is characterised as a shift in DNA sequence that alters a gene's function, single nucleotide polymorphism (SNP) is the most common type of genetic variation, occurring 1 in 1000 base pairs and affecting only one nucleotide. Such monogenic diseases, such as Schizophrenia, Alzheimer's disease, Hutchinson-Gilford Progeria syndrome (HGPS), Proteus syndrome, and Congenital generalised hypertrichosis, are caused by genetic defects (CGH). However, owing to mutations in co-receptors used for infection entry into the body, certain variants have provided immunity against diseases such as HIV infection. This analysis seeks to provide insight into the function of variants found in genes associated with monogenic diseases in order to establish the disease's underlying cause, which may contribute to scientists finding therapeutic ways to cure the disease. Human genetics is one of the most interesting fields of research that leads to a deeper understanding of humans. Human genetics research reveals the causes that cause human diseases. Scientists have recently conquered the mystery of several unusual genetic diseases. Several genes seem to play a role in hereditary disorders. In order to link genetic disorder phenotypes to genotypes, In each human, a certain genetic mutation or genotype is expressed to varying degrees. Those that have a different variant should not have it. Still have the same familiarity of the variant's expression. It's true. It's just not accurate to assume that a mutation is present in any person who carries it. It is often followed by legislation. Penetrance is characterised as a proportion of different individuals having a particular mutation. Total penetrance happens when a person experiences clinical signs of a disease caused by a genetic mutation.

CONCLUSION

There has long been a belief that genetic mutations are often linked to the emergence of a disease, but this situation has improved dramatically thanks to recent developments in genetics and molecular biology [1]. A host of genetic mutations have now been investigated for their involvement with or beneficial function in combating such disorders in specific organisms. The identification of genes implicated in complex and pathological diseases is of greater concern to scientists, physicians, and geneticists. Understanding the genetic mutations that cause tumours, sickle cell anaemia, cystic fibrosis, colour blindness [2], and other illnesses is critical to winning the fight against them. Mining genetic mutations across different genomes has definitely been easier, thanks to a significant improvement in bioinformatics databases and statistical approaches. For the development of any prescription medication to cure genetic or somatic mutations, knowledge of the causes, functions, and curative steps for changing genetic and somatic mutations is absolutely essential [3].

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