



## Health and Disease Effects of Gene Mutations

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### Description

Gene mutations are alterations or changes that occur in the Deoxyribonucleic Acid (DNA) sequence of an individual's genetic material. These mutations can occur in various forms, including point mutations (substitutions, insertions and deletions), chromosomal mutations (inversions, translocations, duplications and deletions) and expansion mutations (repeat expansions). Gene mutations can have significant effects on human health and disease and understanding their impact is high for advancing our knowledge of genetics and developing strategies for disease prevention and treatment.

### Types of gene mutations

Gene mutations can occur in different ways, leading to diverse effects on health and disease. Point mutations involve changes in a single nucleotide base of the DNA sequence and they can result in different outcomes [1]. For example, a substitution mutation can lead to a change in the amino acid sequence of a protein, affecting its structure and function. Insertion and deletion mutations can shift the reading frame of a gene, leading to the production of a non-functional protein.

Chromosomal mutations involve changes in the structure or number of chromosomes. Inversions occur when a segment of a chromosome is reversed, translocations happen when a segment of one chromosome is transferred to another, duplications involve the presence of extra copies of a chromosome segment and deletions result in the loss of a segment of a chromosome. These chromosomal mutations can disrupt gene function and lead to various health conditions [2].

Expansion mutations involve the abnormal expansion of repeat sequences in the DNA. These mutations can cause a wide range of diseases, including neurodegenerative disorders such as Huntington's disease and fragile X syndrome. The severity and age of onset of these diseases are often correlated with the size of the repeat expansion [3,4].

### Role of gene mutations in health

Gene mutations play a vital role in human health. They can act as drivers of evolution and genetic diversity, leading to the emergence of new traits and characteristics in populations. Beneficial gene mutations can confer advantages in certain environments and provide

protection against diseases [5]. For example, mutations that provide resistance to infectious diseases, such as sickle cell trait providing protection against malaria, have been positively selected in some populations.

Genetic variation resulting from gene mutations can also impact individual differences in health and disease. Some gene mutations are associated with improved metabolism, enhanced immunity, or increased resistance to environmental toxins. However, gene mutations can also be associated with increased susceptibility to certain diseases, such as genetic mutations that increase the risk of developing cancer or heart disease [6,7].

### Gene mutations and disease

Gene mutations are often linked to the development of diseases. Pathogenic gene mutations can disrupt normal gene function and lead to various health conditions [8]. For example, mutations in the Breast cancer gene 1 and Breast cancer gene 2, genes are associated with an increased risk of breast and ovarian cancers, while mutations in Cystic Fibrosis Trans membrane Conductance Regulator (CFTR) gene are associated with cystic fibrosis.

Some gene mutations can also result in rare genetic disorders. These disorders are typically caused by mutations in single genes and can affect various aspects of health, including metabolic, developmental and neurological functions. Examples of such disorders include Down syndrome, Duchene muscular dystrophy and Phenylketonuria (PKU).

### Genetic testing and precision medicine

Advancements in genetic testing have made it possible to identify gene mutations associated with specific diseases. Genetic testing can help identify individuals at risk for certain genetic conditions, allowing for early intervention, monitoring and personalized treatment plans. Precision medicine, which involves tailoring medical interventions based on an individual's genetic makeup, holds great promise in improving patient outcomes and preventing diseases associated with gene mutations [8].

### Future directions and challenges

The field of gene mutations and their effects on health and disease is rapidly evolving. Advances in genomic sequencing and gene editing technologies, such as Clustered Regularly Interspaced Short Palindromic Repeats-CRISPR-associated protein 9 (CRISPR-Cas9), are providing unprecedented opportunities for studying gene mutations and their impact on health. With these tools, scientists can better understand the functional consequences of gene mutations, identify novel disease-associated mutations and develop targeted therapies for individuals with genetic disorders [9,10].

### Conclusion

Gene mutations play a vital role in human health and disease. Understanding the different types of gene mutations and their impact on health is essential for advancing the knowledge of genetics and developing effective strategies for disease prevention, diagnosis and treatment. Genetic testing and precision medicine hold great promise in personalized healthcare, but challenges related to ethics, interpretation of genetic data and accessibility need to be addressed.

## References

1. Adamczyk-Poplawska M, Lower M, Piekarowicz A (2011) Deletion of one nucleotide within the homonucleotide tract present in the hsdS gene alters the DNA sequence specificity of type I restriction-modification system NgoAV. *J Bacteriol* 193:6750-6759.
2. McEwen J, Silverman P (1980) Chromosomal mutations of *Escherichia coli* that alter expression of conjugative plasmid functions. *Proc Natl Acad Sci U S A* 77:513-517.
3. Yum K, Wang ET, Kalsotra A (2017) Myotonic dystrophy: Disease repeat range, penetrance, age of onset, and relationship between repeat size and phenotypes. *Curr Opin Genet Dev* 44:30-37.
4. Paulson H (2018) Repeat expansion diseases. *Handb Clin Neurol* 147:105-123.
5. Carter AJR, Nguyen AQ (2011) Antagonistic pleiotropy as a widespread mechanism for the maintenance of polymorphic disease alleles. *BMC Med Genet* 12:1-3.
6. Lapunzina P, López RO, Laguna LR, Miguel PG, Martínez AR, et al. (2014) Impact of NGS in the medical sciences: Genetic syndromes with an increased risk of developing cancer as an example of the use of new technologies. *Genet Mol Biol* 37:241-249.
7. Pfeffer TJ, Pietzsch S, Kleiner DH (2020) Common genetic predisposition for heart failure and cancer. *Herz* 45:632-636.
8. Betancur C, Sakurai T, Buxbaum JD (2009) The emerging role of synaptic cell-adhesion pathways in the pathogenesis of autism spectrum disorders. *Trends Neurosci* 32:402-412.
9. Phillips KA, Trosman JR, Kelley RK, Pletcher MJ, Douglas MP, et al. (2014) Genomic sequencing: assessing the health care system, policy big-data implications. *Health Aff (Millwood)* 33:1246-1253.
10. Dugger SA, Platt A, Goldstein DB (2018) Drug development in the era of precision medicine. *Nat Rev Drug Discov* 17:183-196.