

Understanding Genetic Mutations and their DNA Sequence

Kentro Hayashida*

Department of Cardiology, Keio University School of Medicine, Tokyo, Japan

DESCRIPTION

The genetic instructions can be read out by using the language called DNA. This genetic code, composed of nucleotide sequences, holds the instructions to form and function. However, the code is not immutable; the changes to your DNA sequence known as genetic mutations. This article will discuss about the genetic mutations, exploring their nature, causes, and the profound impact they can have on the life. Deoxyribonucleic Acid (DNA) is a double-stranded molecule that carries the genetic instructions used in the development and functioning of all known living organisms. The structure of DNA resembles a twisted ladder, commonly referred to as the double helix. The rungs of this ladder are made up of pairs of nucleotides; Adenine (A) Thymine (T), Cytosine (C) Guanine (G).

These nucleotide sequences encode the necessary information for the synthesis of proteins, the building blocks of cells. The precise arrangement of these nucleotides forms the genetic code, characteristics and functions of living organisms. Genetic mutations are alterations in the DNA sequence. These mutations can occur in various ways, and their consequences can range from inconsequential to life-altering.

Types of mutations

Point mutations: These involve the replacement of a single nucleotide with another.

Frameshift mutations: These occur when the insertion or deletion of nucleotides disrupts the reading frame of the genetic code. This can lead to a cascade of incorrect amino acid sequences and the production of non-functional proteins.

Insertions and deletions: These involve the addition or removal of nucleotides, potentially causing a frameshift mutation or altering the reading frame.

Spontaneous mutations arise naturally during DNA replication or repair due to errors in the biochemical processes involved. Environmental factors like external influences, such as exposure to radiation, certain chemicals, or mutagenic substances, can induce mutations. Inherited mutations can be passed down

from parents to offspring through the germ cells (sperm and egg cells).

Cystic fibrosis caused by mutations in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene, this disorder leads to the production of thick and sticky mucus, affecting the respiratory and digestive systems. Mutations in genes related to blood clotting can result in hemophilia, a condition where blood doesn't clot properly, leading to excessive bleeding.

Cancer is not solely caused by mutations, many cancers result from the accumulation of genetic mutations that drive uncontrolled cell growth. Genetic factors, including mutations, can contribute to the risk of developing Alzheimer's disease, a neurodegenerative disorder. The role of genetic mutations in cancer, often considered a genetic disease, is characterized by the uncontrolled proliferation of cells. While mutations alone do not guarantee cancer development, certain mutations can disrupt the normal regulatory mechanisms that control cell division, leading to the formation of tumors. Mutations that activate oncogenes can promote cell growth and division, contributing to the development of cancer. Mutations that inactivate tumor suppressor genes remove the normal checks on cell division, also facilitating cancer progression. Skin cancers often exhibit mutational signatures associated with exposure to Ultraviolet (UV) radiation from the sun. Mutations linked to tobacco smoke exposure are observed in lung cancers, highlighting the environmental impact on mutational patterns.

CONCLUSION

Advances in genetic technologies have enabled the identification of specific mutations associated with genetic disorders, susceptibility to diseases, and even responsiveness to certain treatments. Genetic testing plays a crucial role in diagnosing genetic conditions, predicting disease risk, and guiding personalized treatment approaches. Assessing individuals for the presence of mutations associated with recessive genetic disorders, informing decisions about family planning. Identifying genetic abnormalities in developing fetuses to provide early information's about potential health issues.

Correspondence to: Kentro Hayashida, Department of Cardiology, Keio University School of Medicine, Tokyo, Japan, E-mail: verondel@outlook.com

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