

Identifying and Understanding of Disease Genes

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ABOUT THE STUDY

Disease genes are genes that, when mutated or altered, can lead to the development of a wide range of diseases, including genetic disorders, cancer, and various other diseases. These genes may play a crucial role in the development and progression of these diseases, and understanding their functions and mechanisms is crucial for developing effective treatments and cures. Genetic disorders are caused by mutations in one or more genes, which can result in abnormal protein production or function. Some genetic disorders are inherited from one or both parents, while others can occur sporadically due to random mutations. Some examples of genetic disorders include cystic fibrosis, sickle cell anemia, and Huntington's disease. Cancer is a disease that arises due to the abnormal growth and division of cells. It is often caused by mutations in genes that regulate cell growth, division, and death. These genes are known as oncogenes and tumor suppressor genes, respectively. Mutations in these genes can cause uncontrolled cell growth and division, leading to the formation of tumors. Some examples of cancer-associated genes include the BRCA1 and BRCA2 genes, which are associated with an increased risk of breast and ovarian cancer. Other diseases, such as cardiovascular disease, diabetes, and Alzheimer's disease, are multifactorial and involve a combination of genetic and environmental factors. In these diseases, multiple genes may be involved, each contributing to the overall risk and severity of the disease. Once a disease gene has been identified, researchers can investigate its function and develop targeted therapies to treat the disease. For example, in cystic fibrosis, a genetic disorder caused by mutations in the Cystic Fibrosis Transmembrane conductance Regulator (CFTR) gene, researchers have developed drugs that target the defective protein produced by the mutated gene. These drugs can help improve lung function and reduce the risk of infections in individuals with cystic fibrosis.

Identification of disease genes

The identification of disease genes is a complex and challenging process that requires a combination of genetic and bioinformatic approaches. The following are some of the commonly used methods for identifying disease genes.

Genome-Wide Association Studies (GWAS): GWAS is a powerful method used to identify genetic variants associated with complex diseases. GWAS involves comparing the genomes of thousands of individuals with and without a disease to identify genetic variations that are more common in the affected individuals.

This method has been used successfully to identify genetic variants associated with many complex diseases, including diabetes, heart disease, and Alzheimer's disease.

Linkage analysis: Linkage analysis is a method used to identify the location of disease genes on a chromosome. This method involves analyzing the inheritance patterns of genetic markers in families affected by a disease. Linkage analysis can be used to identify disease genes responsible for single-gene disorders.

Candidate gene approach: The candidate gene approach involves selecting genes that are known to be involved in a particular biological process or pathway and testing them for association with a disease. This approach is useful for identifying disease genes responsible for single-gene disorders.

Next-Generation Sequencing (NGS): NGS is a powerful technique that allows the rapid sequencing of large amounts of DNA. This technique has revolutionized the field of genetics and has led to the discovery of many disease genes. NGS can be used to sequence the entire genome or specific regions of interest.

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