

Emerging Concept of Genetically Transitional Disease

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Introduction

Genomic medicine, fueled by advancements in genetics and biotechnology, has revolutionized our understanding of human health and disease. The ability to decode an individual's genetic blueprint has uncovered insights into the genetic basis of various disorders. Among these, a novel and emerging concept has surfaced: genetically transitional disease. This concept challenges traditional disease classifications and offers a more nuanced perspective on how genetics shapes health outcomes. In this article, we will explore the concept of genetically transitional disease, its implications for genomic medicine, examples from various medical fields, and the potential it holds for personalized healthcare. Traditionally, diseases have been classified into two categories: genetic and non-genetic. Genetic diseases are caused by mutations or variations in a person's DNA, while non-genetic diseases are attributed to environmental factors, lifestyle choices, infections, or other non-genetic influences. However, this binary classification does not adequately capture the complexity of disease etiology. Genetically transitional diseases are characterized by a complex interplay between genetic and non-genetic factors in their development and progression. Rather than being solely driven by genetic mutations, these diseases result from a combination of genetic predisposition and environmental influences. The genetic component may increase an individual's susceptibility to the disease or modulate its severity, while non-genetic factors can trigger or exacerbate the condition. While genetics play a significant role in an individual's susceptibility to type 2 diabetes, lifestyle factors such as diet, physical activity, and obesity are also major contributors. Individuals with a genetic predisposition may be more vulnerable to the disease, but environmental factors can tip the balance [1].

Description

Genetic variants can increase the risk of cardiovascular disease, but lifestyle choices such as smoking, diet, and physical inactivity play a substantial role in its development. Additionally, stress and socioeconomic factors can further influence disease outcomes. Alzheimer's disease has a strong genetic component, with specific gene variants increasing the risk. However, environmental factors such as education, cognitive engagement, and vascular health contribute significantly to the disease's onset and progression. Many cancers are considered genetically transitional diseases. Genetic mutations may increase susceptibility to cancer, but lifestyle factors like tobacco use, diet, sun exposure, and environmental toxins play pivotal roles in cancer development. Conditions like depression and schizophrenia have a complex etiology involving genetic predisposition and environmental triggers. Stress, trauma, and childhood experiences can significantly impact the risk and severity of these disorders. The concept of genetically transitional disease underscores the importance of understanding how genetic predisposition

interacts with environmental triggers. Genomic medicine has ushered in a new era of healthcare, where the genetic code of an individual plays a central role in understanding health and disease. In recent years, a concept known as "genetically transitional disease" has emerged, challenging traditional notions of genetic disorders. This paradigm shift in our understanding of genetic conditions acknowledges the dynamic nature of genetics and its impact on health over time [2].

In this article, we will explore the concept of genetically transitional disease, its implications for genomic medicine, and the potential it holds for improving diagnosis, treatment, and prevention of genetic conditions. These are caused by mutations in a single gene and typically follow classical Mendelian inheritance patterns, such as autosomal dominant or recessive. Examples include cystic fibrosis, Huntington's disease, and sickle cell anemia. These conditions involve multiple genetic and environmental factors. They are often characterized by a complex interplay of genetic variants, making them challenging to predict or diagnose. Examples include diabetes, heart disease and many types of cancer. However, recent advances in genomics have revealed that the relationship between genetics and disease is more nuanced than previously thought. The concept of genetically transitional disease has emerged as a result. Genetically transitional disease challenges the notion of a fixed genetic destiny. It acknowledges that genetic information is not static but can change over time, influencing an individual's health and disease risk. This change can occur through various mechanisms. Somatic mutations are genetic changes that occur in non-reproductive cells during an individual's lifetime. These mutations can lead to diseases like cancer, where genetic alterations accumulate in cells, causing uncontrolled growth. Epigenetic changes involve modifications to DNA or associated proteins that can alter gene expression without changing the underlying DNA sequence [3].

Epigenetic modifications can be influenced by environmental factors and lifestyle choices. Mosaicism refers to the presence of genetically distinct cell populations within an individual's body. It can arise from post-zygotic mutations during development and can lead to varying disease manifestations in different tissues. Genetic disease risk can change based on an individual's exposure to environmental factors, such as diet, toxins, infections, and stress. These interactions can modify the expression of disease-associated genes. Cancer is a classic example of a genetically transitional disease. Somatic mutations accumulate over time, driving the development and progression of tumors. Genomic analyses of tumors have led to targeted therapies that exploit these mutations. Conditions like Alzheimer's and Parkinson's disease involve complex gene-environment interactions. Genetic predisposition alone may not determine disease onset, as environmental factors may play a role in triggering or accelerating these conditions. Autoimmune diseases, such as rheumatoid arthritis and multiple sclerosis, have a genetic component but can exhibit dynamic disease courses influenced by environmental factors. Genetic variants contribute to cardiovascular disease risk, but lifestyle factors like diet, exercise, and smoking can significantly impact an individual's risk over time. Dynamic Risk Assessment: Rather than assigning a fixed genetic risk, healthcare providers can adopt a dynamic risk assessment approach that considers the evolving nature of genetic information. Regular genetic testing and monitoring can inform personalized risk management strategies [4].

Recognizing that genetics alone may not predict disease onset, genomic medicine can emphasize preventive interventions that target modifiable risk factors, such as lifestyle changes and early detection. In cases of genetically transitional diseases like cancer, treatments can be tailored based on the evolving genetic profile of the tumor. This approach allows for more effective and personalized therapies. Patients can benefit from a deeper understanding of the dynamic nature of genetic information. Educating individuals about

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the influence of lifestyle choices, environmental exposures and somatic mutations on their health empowers them to make informed decisions. Robust data protection measures are essential to safeguard individuals' sensitive information. Patients must be informed about the potential for dynamic changes in genetic risk and the implications of ongoing genetic testing. Informed consent processes need to be comprehensive and transparent. Access to genetic counseling becomes even more critical in the context of genetically transitional disease. Patients need guidance on interpreting evolving genetic information and its relevance to their health. Ensuring equitable access to genomic medicine, including ongoing genetic testing and counseling, is essential to prevent disparities in healthcare. Long-term, large-scale studies will help unravel the dynamic nature of genetics and its impact on health. These studies can inform disease prevention and treatment strategies. Technological advancements will lead to more sensitive and precise diagnostic tools for monitoring genetic changes over time [5].

Conclusion

Genetically transitional disease is a groundbreaking concept in genomic medicine that challenges traditional notions of fixed genetic destinies. It recognizes the dynamic nature of genetic information and its impact on health over time, driven by somatic mutations, epigenetic modifications, mosaicism, and gene-environment interactions. This concept has far-reaching implications for disease risk assessment, prevention, treatment, and patient education. While it presents challenges related to privacy, informed consent, and equity, genetically transitional disease opens the door to innovative approaches in genomics and healthcare. As research and technology continue to advance, our understanding of the dynamic interplay between genetics and disease will deepen, leading to more personalized and effective healthcare interventions. Artificial intelligence and machine learning algorithms will play a crucial role in analyzing and interpreting the complex, evolving genetic data of individuals. Advancements in regenerative medicine may offer the potential to reverse

or repair somatic mutations responsible for genetically transitional diseases. Genomic medicine will move towards creating personalized health plans that adapt to an individual's changing genetic risk and health status. Genetically transitional disease is a new concept that bridges the gap between these two categories.

Acknowledgement

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Conflict of Interest

None.

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