

Genetic Diseases and the Role of Genetics in Disease Susceptibility

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Description

Genetic diseases are disorders caused by abnormalities in an individual's genetic material, typically inherited from parents or arising from spontaneous mutations. These conditions can affect various aspects of health and life expectancy, presenting unique challenges for patients, families, and healthcare providers. This article explores several genetic diseases-cystic fibrosis, sickle cell disease, and Huntington's disease-and examines the role of genetics in disease susceptibility. Cystic Fibrosis (CF) is a genetic disorder characterized by the buildup of thick, sticky mucus in the lungs and digestive system. It is caused by mutations in the CFTR gene, which regulates the movement of salt and water in and out of cells. Mutations in this gene lead to the production of abnormally thick mucus, impairing lung function and digestion.CF is inherited in an autosomal recessive pattern, meaning a person must inherit two copies of the defective CFTR gene (one from each parent) to develop the disease. Carriers, who have one normal and one mutated CFTR gene, typically do not show symptoms but can pass the mutated gene to their children. Symptoms of CF vary but often include persistent cough with thick mucus, recurrent lung infections, digestive problems, and poor growth. Management involves medications to thin mucus, antibiotics for infections, nutritional therapies, and sometimes lung transplantation for severe cases. Sickle Cell Disease (SCD) is a group of inherited red blood cell disorders. It results from a mutation in the HBB gene, which encodes haemoglobin-the protein that carries oxygen in red blood cells. In SCD, haemoglobin molecules form stiff rods, causing red blood cells to become sickle-shaped and sticky. SCD is inherited in an autosomal recessive pattern. A person must inherit two copies of the mutated HBB gene (one from each parent) to develop the disease. Carriers, like CF, usually do not have symptoms but can pass the mutated gene to their children. Symptoms of SCD include episodes of severe pain (called sickle cell crises), anaemia, increased risk of infections, and organ damage. Management focuses on preventing and managing complications, including pain management, hydration, blood transfusions, and medications to reduce symptoms and complications. Huntington's Disease (HD) is a progressive brain disorder caused by a mutation in the HTT gene, which provides instructions for making a protein called huntingtin.

The mutated protein leads to gradual nerve cell deterioration in the brain, affecting movement, cognition, and behaviour. HD is inherited in an autosomal dominant pattern, meaning a person needs only one copy of the mutated HTT gene (inherited from one affected parent) to develop the disease. Each child of an affected parent has a 50% chance of inheriting the mutated gene. Symptoms of HD typically appear in adulthood and worsen over time, including involuntary movements (chorea), cognitive decline, mood changes, and eventually loss of ability to function independently. Management involves medications to manage symptoms, psychotherapy, and supportive care to improve quality of life. Genetics plays a crucial role in determining an individual's susceptibility to various diseases, including genetic disorders and complex diseases influenced by multiple genes and environmental factors. Genetic testing and counselling can help individuals and families understand their risk of developing certain genetic diseases, allowing for informed decision-making about family planning and health management. Knowledge of genetic mutations can guide personalized medicine approaches, tailoring treatments to individual genetic profiles for improved efficacy and reduced side effects. Genetic studies contribute to advancements in disease understanding, facilitating the development of new therapies, biomarkers, and preventive strategies. In conclusion, genetic diseases such as cystic fibrosis, sickle cell disease, and Huntington's disease highlight the intricate relationship between genetics and health. Advances in genetic research and technology continue to expand our understanding of disease mechanisms, genetic predispositions, and personalized treatment approaches. By integrating genetic knowledge into healthcare practice and policy, we can enhance disease prevention, diagnosis, and management, ultimately improving outcomes and quality of life for individuals affected by genetic disorders worldwide.

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Conflict of Interest

The author declares there is no conflict of interest in publishing this article.

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