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**GLOBAL DISTRIBUTION OF GENETIC DISEASES BY GEOGRAPHIC
REGIONS AND PREVENTIVE STRATEGIES**

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Abstract Genetic diseases constitute a significant global health burden, affecting millions of individuals worldwide. Their distribution varies considerably across geographic regions due to differences in genetic background, population structure, consanguinity rates, environmental factors, and access to healthcare services. This review analyzes the global distribution of genetic diseases by regions and discusses current preventive strategies, including genetic screening, counseling, and public health interventions. Understanding regional epidemiological patterns is essential for effective prevention, early diagnosis, and reduction of disease-related morbidity and mortality.

Keywords Genetic diseases; global distribution; epidemiology; regional prevalence; genetic screening; prevention

Introduction

Genetic diseases are disorders caused by abnormalities in genes or chromosomes and may be inherited or arise from de novo mutations. These conditions include single-gene disorders, chromosomal abnormalities, and multifactorial genetic diseases. Collectively, genetic diseases account for a substantial proportion of congenital anomalies, childhood morbidity, and chronic disability.

The prevalence and spectrum of genetic diseases vary markedly across regions of the world. Factors such as population genetics, migration patterns, consanguinity, and healthcare infrastructure play a major role in shaping regional distribution. This article reviews the global occurrence of genetic diseases by geographic regions and outlines key preventive strategies in line with American and European medical journal standards.[1,2,3]

Global Distribution of Genetic Diseases

North America

In North America, advanced diagnostic technologies and widespread access to genetic testing have led to improved detection and reporting of genetic diseases. Conditions such as cystic fibrosis, sickle cell disease, spinal muscular atrophy, and hereditary cancer syndromes are relatively well-characterized. Comprehensive newborn screening programs significantly contribute to early diagnosis and secondary prevention.

Europe

Europe exhibits regional variability in genetic disease prevalence. Founder effects have resulted in higher frequencies of specific disorders in certain populations, such as Tay–Sachs disease, familial Mediterranean fever, and Huntington disease. European



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countries benefit from coordinated rare disease networks, population registries, and standardized screening programs that enhance epidemiological surveillance.

Asia

Asia demonstrates considerable heterogeneity in the distribution of genetic diseases due to large population size and genetic diversity. Hemoglobinopathies, such as thalassemia, are highly prevalent in South and Southeast Asia. In some regions, limited access to genetic diagnostics leads to underestimation of disease prevalence. Rapid expansion of genomic medicine is improving detection rates.

Africa

Genetic disease data from Africa remain limited, largely due to insufficient diagnostic infrastructure. However, disorders such as sickle cell disease and glucose-6-phosphate dehydrogenase (G6PD) deficiency are highly prevalent. Many inherited metabolic and chromosomal disorders are believed to be underdiagnosed rather than rare.

Middle East

The Middle East shows a relatively high prevalence of autosomal recessive genetic diseases, primarily due to consanguineous marriages. Inherited metabolic disorders, congenital malformation syndromes, and rare genetic conditions are more frequently observed. National premarital and newborn screening programs have been introduced in several countries to reduce disease burden.

Latin America

In Latin America, the distribution of genetic diseases varies widely among countries. Founder mutations, ethnic diversity, and healthcare disparities influence prevalence patterns. Limited access to genetic services contributes to delayed diagnosis, although regional initiatives are improving awareness and screening.[4,5,6]

Factors Influencing Regional Variation

Key determinants of regional differences in genetic disease prevalence include:

- Genetic diversity and founder effects
- Consanguinity and marriage patterns
- Population size and migration
- Environmental influences
- Availability of laboratory diagnostics and screening programs

Prevention and Prophylaxis of Genetic Diseases

Primary Prevention

Primary prevention aims to reduce the occurrence of genetic diseases and includes:

- Carrier screening in high-risk populations
- Premarital and preconception genetic counseling
- Public education on genetic risk factors

Secondary Prevention

Secondary prevention focuses on early detection:

- Newborn screening programs
- Prenatal screening and diagnostic testing
- Early laboratory and genetic diagnosis



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Tertiary Prevention

Tertiary prevention aims to reduce complications and improve quality of life through:

Early treatment and long-term follow-up

Multidisciplinary care

Rehabilitation and supportive therapies

Public Health and Ethical Considerations

Preventive strategies must be implemented with consideration of ethical principles, including informed consent, confidentiality, and equitable access to care. Global disparities in access to genetic services highlight the need for international collaboration, capacity building, and policy development.[7,8]

Conclusion

Genetic diseases show significant regional variation worldwide, influenced by genetic, demographic, and healthcare-related factors. Strengthening preventive strategies through genetic screening, counseling, and public health programs is essential to reduce the global burden of genetic diseases. Continued investment in genomic medicine and international cooperation will play a critical role in improving prevention, early diagnosis, and patient outcomes.

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