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## GLOBAL DISTRIBUTION OF ORPHAN DISEASES BY GEOGRAPHIC REGIONS AND PREVENTIVE STRATEGIES

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**Abstract** Orphan diseases, also known as rare diseases, affect a small proportion of the population individually but collectively impose a substantial global health burden. Their prevalence varies significantly across geographic regions due to genetic, environmental, demographic, and healthcare-related factors. This review analyzes the global distribution of orphan diseases by regions and discusses current preventive strategies, including early diagnosis, genetic counseling, and public health interventions. Understanding regional patterns is essential for improving prevention, diagnosis, and equitable healthcare access worldwide.

**Keywords** Orphan diseases; rare diseases; global distribution; epidemiology; regional prevalence; prevention

### **Introduction**

Orphan diseases are a diverse group of conditions characterized by low prevalence and high clinical complexity. In the United States, a disease is classified as rare if it affects fewer than 200,000 individuals, while the European Union defines rare diseases as those affecting fewer than 1 in 2,000 people. More than 7,000 orphan diseases have been identified globally, affecting over 300 million individuals.

The distribution of orphan diseases is not uniform across regions. Differences in genetic background, consanguinity rates, population structure, environmental exposures, and access to diagnostic services contribute to regional variability. Studying these patterns is essential for developing effective preventive and diagnostic strategies.

### **Global Distribution of Orphan Diseases**

#### **North America**

In North America, particularly the United States and Canada, improved diagnostic infrastructure and widespread use of genetic testing have resulted in higher reported prevalence of orphan diseases. Disorders such as cystic fibrosis, spinal muscular atrophy, and certain inborn errors of metabolism are relatively well-documented. Newborn screening programs play a critical role in early detection.[1,2,3]

#### **Europe**

Europe has a well-established rare disease framework supported by the European Reference Networks (ERNs). Some orphan diseases show higher prevalence in specific populations due to founder effects, such as Tay–Sachs disease in Ashkenazi Jewish



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communities or familial Mediterranean fever in Southern Europe. Comprehensive registries contribute to more accurate epidemiological data.

### **Asia**

Asia exhibits considerable heterogeneity in the distribution of orphan diseases. High population density and genetic diversity contribute to variable prevalence. In some regions, high rates of consanguineous marriages increase the incidence of autosomal recessive orphan diseases. Limited access to advanced laboratory diagnostics leads to underreporting in low-resource settings.

### **Africa**

Data on orphan diseases in Africa remain limited due to insufficient diagnostic infrastructure and lack of national registries. However, certain rare genetic and metabolic disorders are believed to be underdiagnosed rather than absent. Infectious and environmental factors may also contribute to region-specific rare conditions.

### **Middle East**

The Middle East shows a relatively high prevalence of autosomal recessive orphan diseases, largely due to consanguinity. Disorders such as inherited metabolic diseases and rare congenital syndromes are more frequently observed. Several countries have begun implementing national genetic screening and counseling programs.

### **Latin America**

In Latin America, orphan disease prevalence varies widely. Limited healthcare resources, unequal access to genetic testing, and lack of standardized registries contribute to diagnostic delays. Nevertheless, regional initiatives are improving awareness and data collection.[4,5,6]

### **Factors Influencing Regional Distribution**

Key factors affecting regional variation include:

Genetic diversity and founder mutations

Rates of consanguineous marriage

Population size and structure

Availability of laboratory diagnostics

Public health policies and screening programs

### **Prevention and Prophylaxis of Orphan Diseases**

#### **Primary Prevention**

While many orphan diseases are genetic and cannot be entirely prevented, risk reduction strategies include:

Carrier screening in high-risk populations

Avoidance of consanguineous marriages when possible

Public education and awareness programs

#### **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 disease severity and complications through:

Early treatment and long-term follow-up

Multidisciplinary care

Patient registries and specialized reference centers

### Public Health and Ethical Considerations

Global disparities in orphan disease diagnosis raise ethical concerns regarding equity and access to care. International collaboration, data sharing, and harmonization of diagnostic standards are essential. Ethical issues related to genetic testing, informed consent, and data privacy must be addressed within culturally appropriate frameworks.[7,8]

### Conclusion

The global distribution of orphan diseases varies significantly across regions due to genetic, demographic, and healthcare-related factors. Many orphan diseases remain underdiagnosed, particularly in low- and middle-income countries. Strengthening laboratory diagnostic capacity, expanding screening programs, and improving preventive strategies are critical to reducing the global burden of orphan diseases. Coordinated international efforts are essential to ensure equitable care and improved outcomes worldwide.

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