ISSN: 2692-5206
AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

PREVALENCE OF GENETIC DISORDERS IN CENTRAL ASIA OVER THE PAST 10 YEARS

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Relevance of the Study

The relevance of this study lies in its comprehensive examination of the frequency and patterns of genetic disorders in Central Asia—a region where such conditions are often overlooked in public health planning. Due to cultural practices such as consanguineous marriages and limited access to specialized genetic services, the risk of hereditary diseases remains high.

Understanding the prevalence of these conditions over the past decade is crucial for:

- Developing national strategies for early detection and prevention.
- Raising awareness among healthcare providers and policymakers.
- Guiding future research and allocation of healthcare resources.
- Reducing the burden of preventable genetic disorders through timely intervention.

This research offers crucial insights for enhancing genetic healthcare infrastructure, promoting genetic literacy, and ultimately improving the quality of life for individuals and families affected in the region.

Keywords: Genetic disorders, Central Asia, prevalence, consanguinity, hereditary diseases, public health, newborn screening, epidemiology, autosomal recessive, thalassemia, Down syndrome

Introduction

Genetic disorders are diseases that are caused by abnormalities in an individual's DNA. These abnormalities can be inherited from parents or arise de novo during early development. The study of genetic disorders is especially important in regions where consanguineous marriages and limited access to genetic counseling are common. Central Asia—a region comprising countries such as Kazakhstan, Uzbekistan, Kyrgyzstan, Turkmenistan, and Tajikistan—has a unique genetic landscape influenced by ethnic diversity, cultural traditions, and socio-economic factors.

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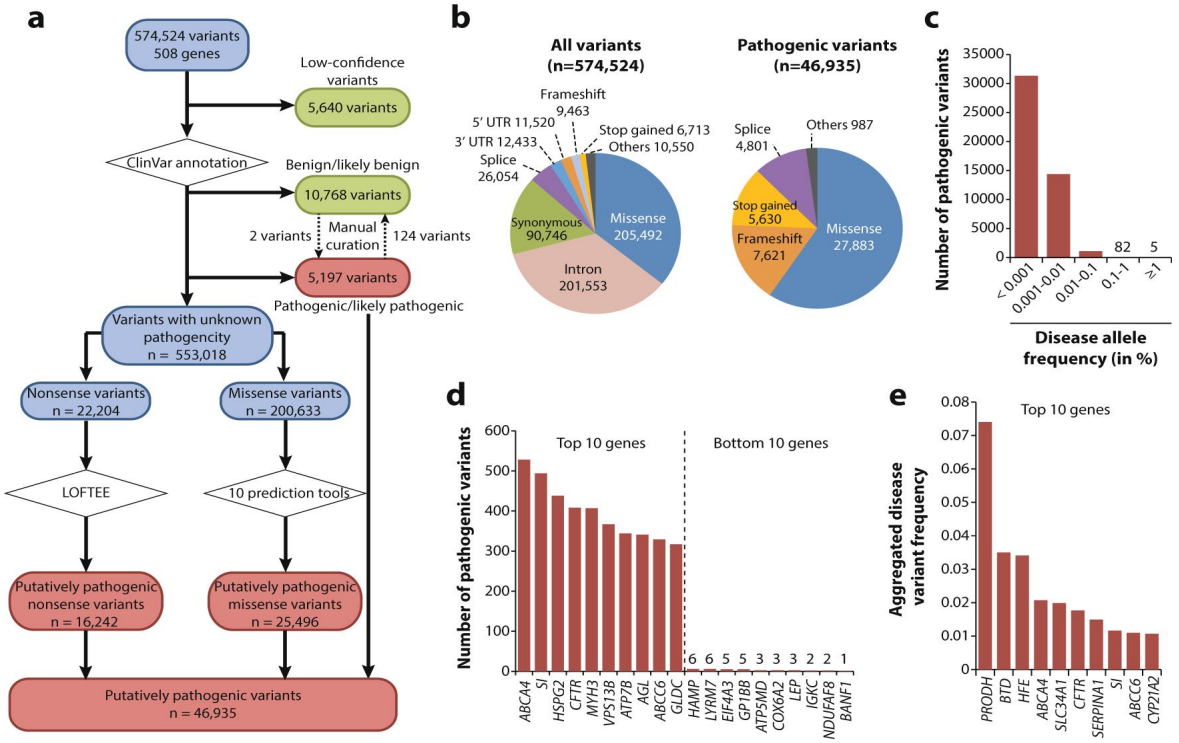
Over the past decade, there has been growing concern regarding the prevalence of genetic diseases in this region. Public health systems are gradually beginning to recognize the need for early diagnosis, screening programs, and preventive strategies. However, limited data and underreporting remain significant challenges. This article aims to analyze the frequency and distribution of genetic disorders in Central Asia from 2014 to 2024, based on available epidemiological data and clinical studies.



Aim of the Study

The primary goal of this study is to investigate the frequency and distribution of genetic disorders in Central Asia over the past 10 years. Specific objectives include:

To identify the most common genetic disorders in Central Asian populations.

To analyze trends in prevalence over the past decade.

To examine contributing factors such as consanguinity, environmental influences, and public health initiatives.

To evaluate the availability and impact of genetic screening and counseling programs in the region.

Materials and Methods

This study is based on a systematic review of available literature, official health statistics, and medical research articles published between 2014 and 2024. Data sources included:

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AMERICAN ACADEMIC PUBLISHER
INTERNATIONAL JOURNAL OF MEDICAL SCIENCES

National healthcare reports from Central Asian ministries of health.

Research databases such as PubMed, Scopus, and WHO regional publications.

Hospital records and clinical case studies from major medical centers in Uzbekistan, Kazakhstan, and Kyrgyzstan.

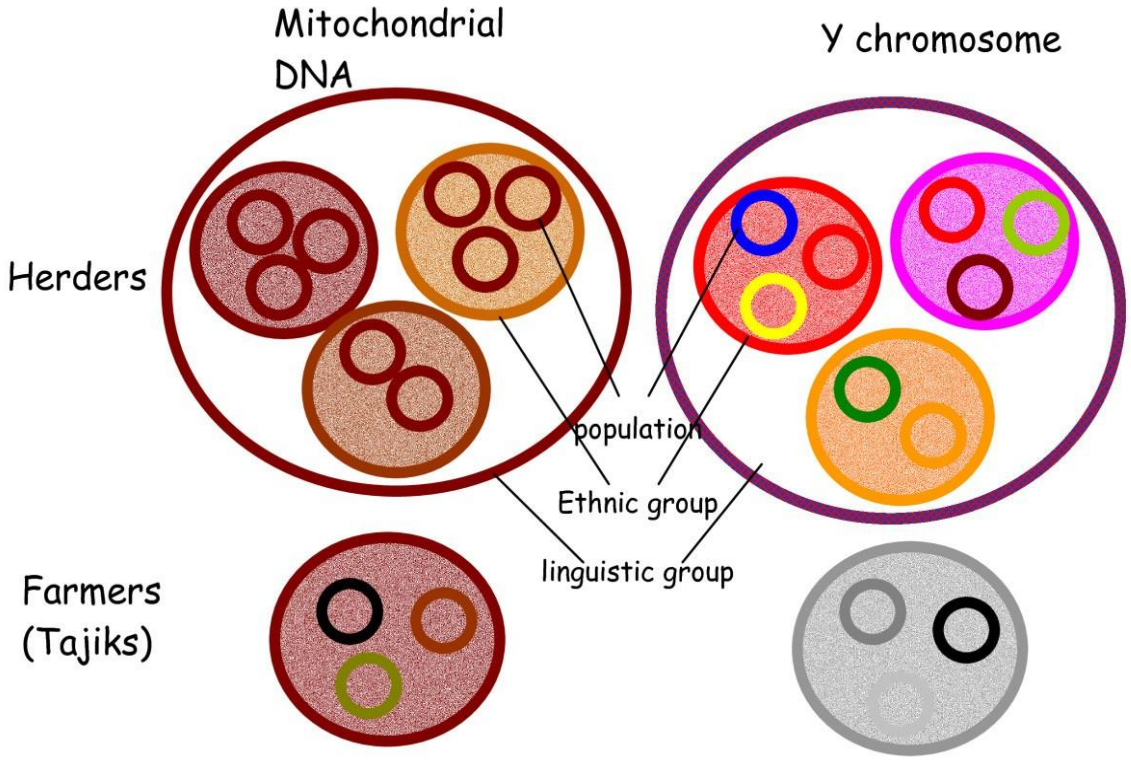
Inclusion criteria:

- Studies and reports that contained epidemiological data on genetic disorders.
- Reports focused specifically on Central Asian populations.

Exclusion criteria:

- Studies unrelated to genetic or hereditary diseases.
- Data lacking specific regional focus or time frame.

Statistical analysis was conducted to determine the frequency of various genetic disorders and assess trends over time. Where possible, prevalence was calculated per 100,000 population.



Results

ISSN: 2692-5206
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The results revealed significant variation in the prevalence of genetic disorders across Central Asian countries. Key findings include:

High prevalence of hemoglobinopathies (e.g., thalassemia and sickle cell disease), particularly in southern regions of Uzbekistan and Tajikistan, with rates ranging from 10 to 30 cases per 100,000 people.

Increased rates of autosomal recessive disorders such as cystic fibrosis, phenylketonuria, and congenital adrenal hyperplasia, especially in rural areas with high rates of consanguineous marriages.

Reported cases of chromosomal disorders, such as Down syndrome, have remained relatively stable but show a slight increase due to improved prenatal diagnostics.

Limited newborn screening programs, with pilot projects introduced in parts of Kazakhstan and Uzbekistan in the last five years, show promising results for early detection.

Genetic counseling services remain underdeveloped, with only a few specialized centers available in capital cities.

Environmental factors, such as exposure to pollutants and radiation (notably in regions near the former Semipalatinsk nuclear test site), may contribute to an increased risk of certain congenital anomalies.

Overall, the average reported prevalence of genetic disorders across Central Asia was estimated to be between 50 and 200 cases per 100,000 population, depending on the region and availability of diagnostics.

Conclusion

This study highlights the growing importance of addressing genetic disorders as a public health concern in Central Asia. Over the past 10 years, the prevalence of various inherited conditions has remained significant, with certain disorders becoming more detectable due to advances in diagnostics. However, challenges such as underreporting, cultural barriers, and insufficient healthcare infrastructure hinder accurate assessment and timely intervention.

To improve outcomes, there is an urgent need for:

- Expansion of newborn screening programs across all Central Asian countries.
- Greater investment in genetic research and public health education.
- Development of accessible genetic counseling and diagnostic services.
- Regional collaboration to build comprehensive genetic disease registries.
- Addressing these gaps will not only reduce the burden of genetic diseases but also improve overall maternal and child health in the region.

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AMERICAN ACADEMIC PUBLISHER
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