REVIEW

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Human Genetics in Kazakhstan: From Population Structure to Precision Medicine

A Transition Toward Population-Specific Genomic Healthcare

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Abstract

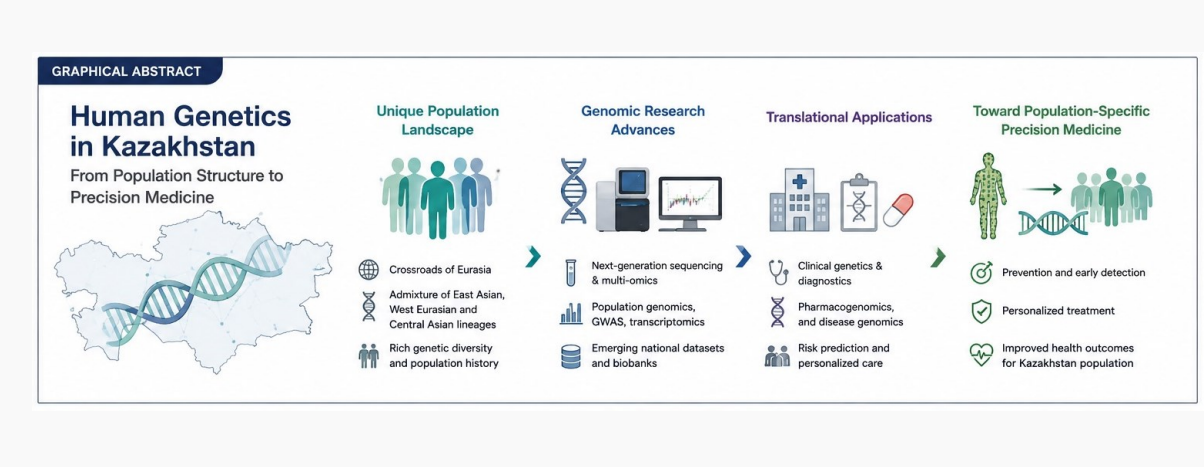
Human genetics in Kazakhstan has undergone substantial transformation over the past three decades, evolving from descriptive population studies toward translational genomics and emerging precision medicine frameworks. Positioned at the crossroads of Eurasia, Kazakhstan represents a genetically heterogeneous population shaped by complex historical migrations, admixture processes, and long-standing interactions between East Asian, West Eurasian, and Central Asian ancestral lineages. This unique genomic architecture provides an important model for studying human diversity, population history, and population-specific disease susceptibility.

This review synthesizes the current state of human genetics research in Kazakhstan across several interconnected domains, including population genomics, medical and clinical genetics, molecular genomics, bioinformatics, and preventive genetics. We further evaluate the evolution of national research infrastructure, implementation of next-generation sequencing technologies, development of genomic datasets, and emergence of translational and personalized medicine initiatives.

Despite significant advances, major challenges remain, including limited large-scale genomic cohorts, insufficient clinical integration of genomic data, regional disparities in access to genetic services, fragmented bioinformatic infrastructure, and underdeveloped ethical and regulatory frameworks. We propose that Kazakhstan is currently transitioning from a population genetics-dominated research system toward a population-specific precision medicine model in which genomic information increasingly informs healthcare decision-making, disease prevention, and individualized therapeutic strategies.

Given its unique admixture-driven genomic heterogeneity and expanding scientific infrastructure, Kazakhstan has the potential to become a strategically important contributor to global population genomics and precision medicine research, particularly for underrepresented Central Eurasian populations.

Keywords: *human genetics; Kazakhstan; genomics; population structure; precision medicine; population-specific genomics; translational genomics; Central Eurasia*



Graphical abstract. From a unique population landscape at the crossroads of Eurasia, through genomic research advances and translational applications, toward population-specific precision medicine for Kazakhstan.

Introduction

Human genetics plays a central role in understanding biological variation, disease susceptibility, and interindividual differences in therapeutic response. Over the past two decades, advances in high-throughput sequencing technologies, genome-wide association studies (GWAS), transcriptomics, and bioinformatics have transformed biomedical research and accelerated the global transition toward precision medicine [1-4]. These developments have enabled the integration of genomic information into disease diagnosis, risk prediction, pharmacogenomics, and personalized therapeutic strategies.

Large-scale international initiatives such as the UK Biobank, the Estonian Genome Project, and national precision medicine programs in China, the United States, and the Middle East have demonstrated the importance of population-specific genomic datasets for improving clinical interpretation and healthcare implementation [5, 6]. However, Central Eurasian populations remain substantially underrepresented in global genomic databases, limiting the transferability of genetic risk prediction models and variant interpretation frameworks [7].

Kazakhstan occupies a strategically important geographic and demographic position in Central Eurasia. The population structure of Kazakhstan reflects centuries of migration, admixture, and population interaction between East Asian, West Eurasian, Turkic, Mongolian, and indigenous Central Asian groups [8-10]. This admixture-driven genomic heterogeneity makes Kazakhstan particularly valuable for studying human evolutionary history, population-specific allele frequency architecture, and the genetic basis of complex diseases.

Human genetics research in Kazakhstan has evolved from Soviet-era cytogenetics and anthropological genetics toward modern molecular genomics and translational medicine [11]. Following independence, the country experienced substantial scientific restructuring, modernization of biomedical infrastructure, and expansion of international collaboration. More recently, the implementation of next-generation sequencing (NGS), whole-genome sequencing (WGS), pharmacogenomics, multi-omics technologies, and computational genomics has facilitated the emergence of precision medicine initiatives within the country [12-15].

This review aims to provide a comprehensive overview of the development of human genetics in Kazakhstan, with particular emphasis on the transition from descriptive population genetics toward clinically actionable genomic medicine. We discuss the major research domains, emerging genomic infrastructure, current limitations, ethical and regulatory considerations, and future strategic directions required for establishing a nationally integrated precision medicine ecosystem.

Materials and Methods

Literature search strategy

A systematic literature search was conducted using PubMed, Scopus, Web of Science, and Google Scholar for studies published up to December 2024. Search terms included combinations of “Kazakhstan” AND “human genetics”, “population genetics”, “medical genetics”, “genomics”, “precision medicine”, “pharmacogenomics”, “hereditary diseases”, “bioinformatics”, and “genomic medicine”. Additional references were identified through citation tracking and manual screening of bibliographies from relevant review articles and primary studies.

Inclusion and exclusion criteria

Studies were included if they were peer-reviewed scientific articles; involved human populations from Kazakhstan; addressed population genetics, clinical genetics, molecular genomics, public health genetics, or bioinformatics; and were published in English or Russian with sufficient methodological detail. The following were excluded:

- studies focused exclusively on animal or plant genetics;
- conference abstracts without full-text availability;
- publications lacking methodological transparency or reproducibility;
- duplicate datasets or redundant analyses.

Data extraction and qualitative analysis

Data were extracted regarding study design, population characteristics, genomic methodologies, sequencing technologies, analytical approaches, and major scientific findings. The identified studies were grouped into thematic categories, including population genomics, medical genetics, molecular and translational genomics, bioinformatics, and preventive genetics. A qualitative synthesis approach was applied to evaluate developmental trends, translational relevance, and emerging national priorities in human genetics research.

Evolution of Human Genetics Research in Kazakhstan

Foundational and Soviet-era genetics

Human genetics research in Kazakhstan originated within the framework of Soviet biomedical science and was primarily focused on cytogenetics, hereditary disease characterization, population anthropology, and family-based genetic analyses. Early investigations emphasized phenotypic variation, demographic genetics, and environmental influences on hereditary disorders. Foundational contributions by R. I. Bersimbayev and colleagues established important institutional and scientific frameworks for the subsequent development of molecular and human genetics research in the country, including long-term work on environmental genotoxicity in exposed populations [11, 25].

Although these studies established important methodological and institutional foundations, they were limited by the absence of high-resolution molecular technologies. During this period, genetics research was largely descriptive and epidemiological, with restricted access to advanced genomic methodologies that later became standard in international biomedical science.

Post-independence scientific transition

Following independence in 1991, Kazakhstan underwent substantial restructuring of its scientific and healthcare systems. This period was characterized by modernization of research infrastructure, expansion of international scientific collaboration, introduction of molecular genetic technologies, development of academic genetics programs, and establishment of specialized biomedical research centers. The adoption of polymerase chain reaction (PCR)-based diagnostics, molecular cytogenetics,

and early sequencing approaches facilitated the transition from classical genetics toward molecular medicine.

Emergence of the genomic era

Since approximately 2010, Kazakhstan has entered an increasingly genomics-oriented phase characterized by implementation of next-generation sequencing (NGS), genome-wide association studies, transcriptomic and epigenomic profiling, computational genomics, and multi-omics integration [12-15].

This transition has enabled the emergence of population-scale genomic initiatives, rare disease sequencing programs, pharmacogenomics studies, and early precision medicine frameworks. Contemporary human genetics research in Kazakhstan increasingly emphasizes clinically actionable genomics, disease mechanisms, and population-specific variant interpretation (Figure 1).

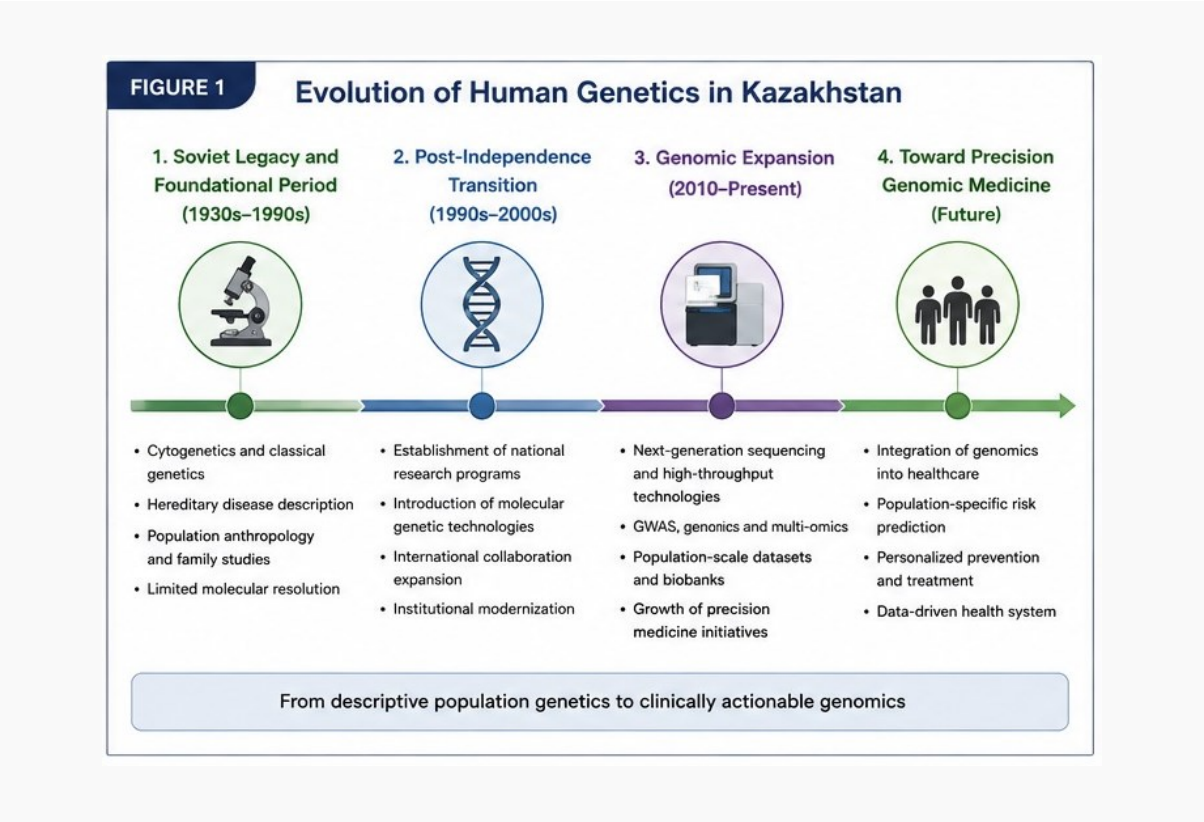


Fig 1. Evolution of human genetics in Kazakhstan. Four developmental periods are shown: the Soviet legacy and foundational period (1930s-1990s), the post-independence transition (1990s-2000s), genomic expansion (2010-present), and the emerging move toward precision genomic medicine, tracing a trajectory from descriptive population genetics to clinically actionable genomics.

Major Research Domains in Human Genetics in Kazakhstan

Population genomics and anthropological genetics

Population genomics remains one of the most extensively developed areas of human genetics research in Kazakhstan. Studies involving mitochondrial DNA, Y-chromosome haplogroups, Y-STR markers, autosomal markers, and genome-wide polymorphism analysis have demonstrated substantial admixture between East Eurasian and West Eurasian ancestral components, and have resolved fine-scale genetic structure among Kazakh tribes and zhuz [8-10, 16, 17].

These investigations have contributed to the reconstruction of migration patterns across Central Eurasia and provided insight into historical population continuity, nomadic migration dynamics, and

regional demographic structure. Importantly, population-specific allele frequency data generated from these studies provide an essential foundation for clinical genomics and genetic risk interpretation in Kazakh populations. Given the underrepresentation of Central Asian populations in global genomic databases, these datasets have increasing translational relevance for precision medicine initiatives.

Medical and clinical genetics

Medical genetics in Kazakhstan has expanded substantially in response to the growing burden of congenital anomalies, inherited metabolic disorders, hereditary cancers, and cardiovascular diseases. Current clinical genetics services include genetic counseling, prenatal diagnostics, cytogenetic analysis, molecular genetic testing, and limited implementation of genomic sequencing technologies. Work by G. S. Svyatova and colleagues contributed substantially to the development of clinical genetics infrastructure, genetic-demographic characterization of Kazakh populations, and genotype-phenotype association studies within the national healthcare system [18, 19].

However, clinical genetics infrastructure remains concentrated primarily in major urban centers, including Astana and Almaty. Regional disparities in access to specialized diagnostics and genetic counseling continue to represent a significant challenge for equitable healthcare delivery.

Molecular genomics and precision medicine

The introduction of high-throughput sequencing technologies has transformed biomedical research capacity in Kazakhstan. Current molecular genomics studies focus on disease-associated genetic variants in local populations, pharmacogenomic determinants of drug response, cancer genomics and transcriptomics, cardiovascular genomics, and rare disease gene discovery [12-15, 20, 21, 24, 26]. Whole-genome and whole-exome sequencing have been applied to early-onset epilepsy, osteogenesis imperfecta, and solid organ transplantation, while genetic risk factors for intracranial aneurysm and high-risk HPV genotype distribution have been characterized in Kazakhstani cohorts.

These efforts reflect a broader transition from descriptive genetics toward mechanism-driven and clinically actionable genomics. Population-specific genomic datasets are increasingly recognized as essential for accurate variant interpretation, polygenic risk assessment, pharmacogenomic implementation, and genotype-guided therapeutic decision-making. Emerging precision medicine initiatives in Kazakhstan increasingly integrate genomic, clinical, and bioinformatic data to support individualized healthcare strategies.

Bioinformatics and computational genomics

The rapid expansion of genomic technologies has increased the importance of bioinformatics and computational genomics in Kazakhstan. Bioinformatic approaches are now essential for NGS data processing, variant annotation, transcriptomic analysis, genomic database development, and integration of multi-omics datasets. National capacity in this area has been demonstrated in whole-genome analyses of drug-resistant *Mycobacterium tuberculosis* and in population-scale human genome variant analysis [13, 22, 23].

The development of national genomic resources and biobank-associated data infrastructure remains an important strategic priority. However, shortages of trained bioinformaticians, computational infrastructure limitations, and fragmented data integration continue to constrain large-scale genomic implementation.

Preventive and public health genetics

Preventive genetics represents an emerging component of healthcare modernization in Kazakhstan. Current applications include newborn screening programs, carrier screening for inherited disorders, prenatal risk assessment, and early detection strategies for non-communicable diseases such as prediabetes and cardiovascular conditions [19].

Although pilot initiatives have demonstrated clinical utility, implementation remains fragmented and insufficiently integrated into national public health systems. Expansion of preventive genomics will require coordinated healthcare policy, standardized genomic guidelines, and population-level genomic literacy programs.

Current Stage of Development: A Transitional Genomic Ecosystem

Human genetics in Kazakhstan currently represents a transitional system situated between classical population genetics and healthcare-integrated precision medicine. Key strengths of the national system include:

- rapidly expanding molecular genetics infrastructure;
- increasing implementation of NGS technologies;
- growing international scientific collaboration;
- emergence of population-specific genomic datasets;
- development of translational genomics initiatives.

At the same time, major limitations persist:

- limited large-scale genomic cohorts and GWAS studies;
- fragmented genomic and clinical data integration;
- insufficient bioinformatic capacity;
- shortage of clinical geneticists and genomic specialists;
- uneven regional access to genetic services;
- underdeveloped national genomic governance systems.

This duality reflects the broader transition from research-centered genetics toward clinically integrated genomic healthcare.

Ethical, Legal, and Social Implications

The expansion of genomic medicine in Kazakhstan introduces important ethical, legal, and social challenges. Major considerations include informed consent in genomic sequencing studies; privacy and protection of genetic information; governance of biobanks and genomic databases; management of incidental findings; equitable access to genomic medicine; and responsible cross-border genomic data sharing [27].

In addition, the development of population-scale genomic resources raises questions regarding genomic sovereignty and long-term stewardship of genetic data derived from underrepresented Eurasian populations. Although Kazakhstan is progressively aligning with international ethical standards, comprehensive national legislation regulating genomic medicine, biobanking, and clinical sequencing remains under development.

Challenges and Future Strategic Directions

Key challenges

Several major barriers currently limit the implementation of precision medicine in Kazakhstan: insufficient funding for large-scale genomic initiatives; limited integration between research institutions and clinical systems; absence of nationally coordinated genomic databases; workforce shortages in genetics and bioinformatics; low public awareness of genomic medicine; and limited interoperability of healthcare data systems.

Strategic priorities

Future development of precision medicine in Kazakhstan should prioritize establishment of a national genomics program; development of integrated biobank and genomic database infrastructure; expansion of clinical genetics services; implementation of standardized genomic healthcare policies; workforce

training in genomics, bioinformatics, and genetic counseling; and strengthening of international multi-center collaboration.

Kazakhstan as a Population-Specific Precision Medicine Model

Kazakhstan represents a potentially important model for population-specific precision medicine due to its unique admixture-driven genomic architecture and rapidly evolving biomedical infrastructure. The current developmental trajectory can be conceptualized as a continuum: population structure → molecular genomics → bioinformatic integration → clinical translation → precision healthcare (Figure 2).

This framework reflects the transition from descriptive ancestry-based studies toward clinically actionable genomic interpretation and individualized healthcare implementation. Such a model may be particularly valuable for ethnically heterogeneous and historically underrepresented populations, where population-specific genomic reference data are essential for accurate risk prediction and therapeutic optimization [4, 7].

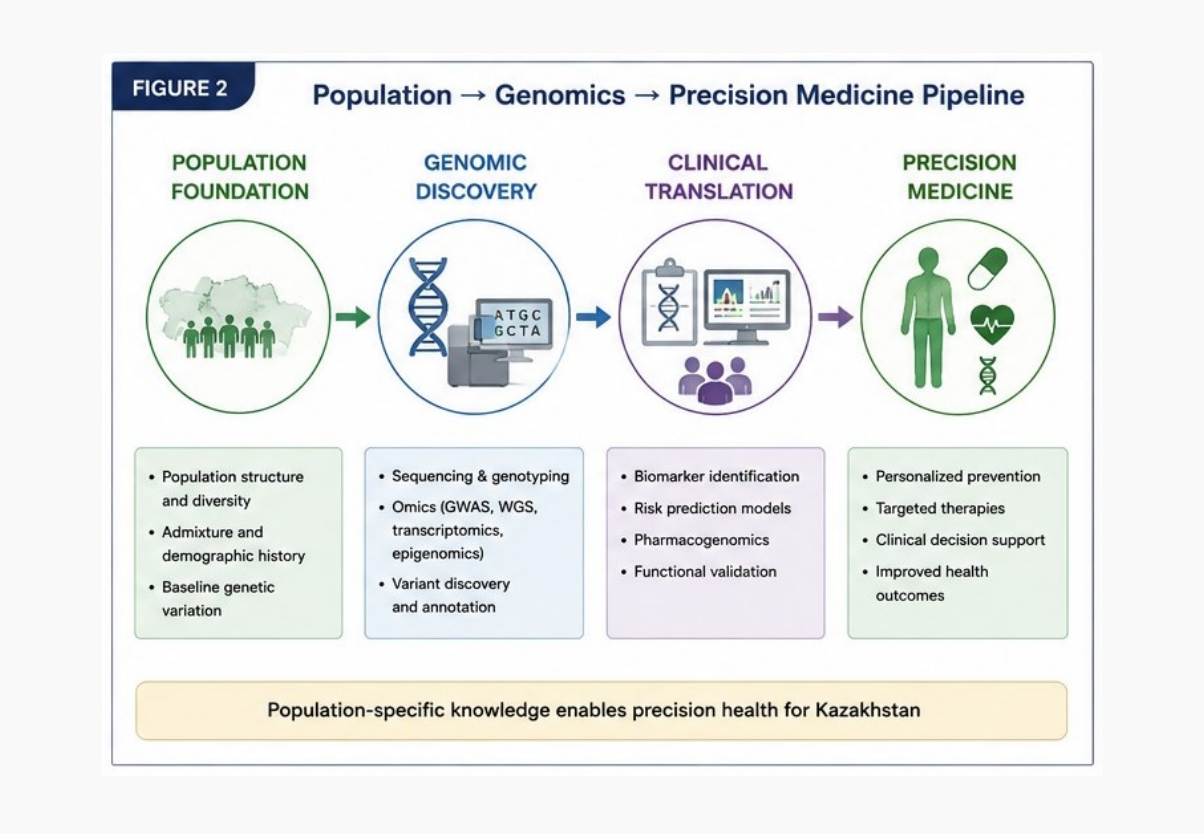


Fig 2. The population to genomics to precision medicine pipeline. Four linked stages - population foundation, genomic discovery, clinical translation, and precision medicine - show how population-specific knowledge enables precision health for Kazakhstan.

Conclusions

Human genetics in Kazakhstan has evolved from classical population and anthropological studies into an emerging ecosystem of translational genomics and precision medicine. While population genetics remains a dominant scientific domain, increasing implementation of high-throughput sequencing, bioinformatics, and clinical genomics is accelerating the transition toward healthcare-integrated genomic medicine.

Sustained progress will depend on the development of national genomic infrastructure, expansion of workforce capacity, establishment of integrated genomic databases, strengthening of ethical governance, and broader incorporation of genomic technologies into healthcare systems.

Given its unique position at the intersection of Eurasian population diversity and emerging genomic science, Kazakhstan has significant potential to contribute to global population genomics, translational medicine, and precision healthcare research, particularly for historically underrepresented Central Asian populations.

Ethics Approval and Consent

Not applicable. This review is based exclusively on previously published literature and did not involve new studies of human participants or animals.

Data Availability Statement

No new datasets were generated or analysed. All data discussed in this review are available in the cited publications.

Author Contributions

Conceptualization: A.R.A.; Methodology: A.R.A.; Investigation: A.R.A.; Writing - original draft: A.R.A.; Writing - review & editing: A.R.A. The author has read and agreed to the published version of the manuscript.

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Competing Interests

The author has declared that no competing interests exist.

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