



METHODS OF STUDYING THE INFLUENCE OF ENVIRONMENTAL FACTORS ON HEREDITARY GENETIC DISEASES

Djalilova Gulchekhra

Rasulova Nilufar

Mukhamedova Nigora

Tashkent Pediatric Medical Institute, Uzbekistan

<https://doi.org/10.5281/zenodo.14916997>

ARTICLE INFO

Received: 18th February 2025

Accepted: 23rd February 2025

Online: 24th February 2025

KEYWORDS

Hereditary genetic diseases, environmental factors, gene-environment interaction, GWAS, epigenetics, epidemiology, twin studies, genetic susceptibility.

ABSTRACT

Hereditary genetic diseases are caused by inherited mutations, but environmental factors can significantly influence their onset, severity, and progression. Understanding the interplay between genetics and environmental exposures is crucial for developing preventive strategies and personalized treatments. This article discusses various methods used to study the impact of environmental factors on genetic diseases, including epidemiological studies, twin studies, genome-wide association studies (GWAS), epigenetic research, and animal models. Each method provides unique insights into gene-environment interactions, contributing to the broader understanding of disease mechanisms.

МЕТОДЫ ИЗУЧЕНИЯ ВЛИЯНИЯ ФАКТОРОВ СРЕДЫ НА НАСЛЕДСТВЕННЫЕ ГЕНЕТИЧЕСКИЕ ЗАБОЛЕВАНИЯ

Джалилова Г.А.

Расулова Н.Ф.

Мухамедова Н.С.

Ташкентский педиатрический медицинский институт, Узбекистан

<https://doi.org/10.5281/zenodo.14916997>

ARTICLE INFO

Received: 18th February 2025

Accepted: 23rd February 2025

Online: 24th February 2025

KEYWORDS

Наследственные генетические заболевания, факторы окружающей среды, взаимодействие генов и среды, GWAS, эпигенетика, эпидемиология, исследования близнецов, генетическая предрасположенность.

ABSTRACT

Наследственные генетические заболевания вызваны наследственными мутациями, но факторы окружающей среды могут существенно влиять на их возникновение, тяжесть и прогрессирование. Понимание взаимодействия между генетикой и воздействием окружающей среды имеет решающее значение для разработки профилактических стратегий и персонализированного лечения. В этой статье обсуждаются различные методы, используемые для изучения влияния факторов окружающей среды на генетические заболевания, включая эпидемиологические исследования, исследования близнецов, полногеномные исследования ассоциаций (GWAS), эпигенетические



исследования и модели на животных. Каждый метод дает уникальную информацию о взаимодействии генов и окружающей среды, способствуя более широкому пониманию механизмов заболеваний.

IRSIY GENETIK KASALLIKLARGA EKOLOGIK FAKTORLARNI TASIRINI O'RGANISH USLUBLARI

Jalilova G.A.

Rasulova N.F.

Muxamedova N.S.

Toshkent pediatriya tibbiyot instituti, O'zbekiston

<https://doi.org/10.5281/zenodo.14916997>

ARTICLE INFO

Received: 18th February 2025

Accepted: 23rd February 2025

Online: 24th February 2025

KEYWORDS

Irsiy genetik kasalliklar, atrof-muhit omillari, gen va atrof-muhitning o'zaro ta'siri, GWAS, epigenetika, epidemiologiya, egizaklarni o'rganish, genetik moyillik.

ABSTRACT

Irsiy genetik kasalliklar irsiy mutatsiyalar tufayli yuzaga keladi, ammo atrof-muhit omillari ularning paydo bo'lishi, zo'ravonligi va rivojlanishiga sezilarli ta'sir ko'rsatishi mumkin. Genetika va atrof-muhitga ta'sir qilish o'rtasidagi o'zaro ta'sirni tushunish profilaktika strategiyalari va shaxsiylashtirilgan davolanishni ishlab chiqish uchun juda muhimdir. Ushbu maqolada genetik kasalliklarga atrof-muhit omillarining ta'sirini o'rganish uchun ishlatiladigan turli usullar, jumladan epidemiologik tadqiqotlar, egizak tadqiqotlar, genom bo'yicha assotsiatsiyalar (GWAS), epigenetik tadqiqotlar va hayvonlar modellari muhokama qilinadi. Har bir usul genlar va atrof-muhit o'rtasidagi o'zaro ta'sirlar haqida noyob ma'lumot beradi, bu kasallik mexanizmlarini kengroq tushunishga yordam beradi.

Introduction. Genetic diseases result from inherited mutations that alter normal biological functions. However, environmental factors such as diet, pollution, radiation, and lifestyle choices can influence disease expression and progression. Understanding these interactions is crucial for improving disease prediction, prevention, and treatment (Lander et al., 2001). Researchers employ various methods to study gene-environment interactions, ranging from large-scale epidemiological surveys to molecular-level genetic analyses. This article explores the primary methods used to investigate the influence of environmental factors on hereditary genetic diseases.

Methods of Studying Gene-Environment Interactions

1. Epidemiological Studies

Epidemiological research is essential in identifying environmental risk factors associated with genetic diseases. These studies involve analyzing large populations to detect correlations between environmental exposures and disease prevalence. Case-control studies, cohort studies, and population-based registries provide crucial data for understanding disease etiology (Brenner et al., 2017).



2. Twin and Family Studies

Twin studies compare monozygotic (identical) and dizygotic (fraternal) twins to assess the relative contributions of genetic and environmental factors to disease development. If a disease shows higher concordance among identical twins than fraternal twins, genetic factors play a significant role. However, if both twin types show similar concordance rates, environmental factors are likely more influential (Boomsma et al., 2002).

3. Genome-Wide Association Studies (GWAS)

GWAS is a powerful method that examines genetic variations across large populations to identify genetic susceptibility to diseases. These studies correlate specific genetic markers with disease occurrence, helping to identify genetic variants that may interact with environmental factors. For example, GWAS has identified genetic predispositions for diseases such as asthma, diabetes, and certain cancers that are exacerbated by environmental exposures (Visscher et al., 2017).

4. Epigenetic Research

Epigenetics examines changes in gene expression that occur without altering the DNA sequence. Environmental factors such as toxins, diet, and stress can modify epigenetic markers like DNA methylation and histone modifications, influencing disease risk. Research on epigenetic changes provides insight into how external factors can activate or suppress genetic diseases (Feil & Fraga, 2012).

5. Animal Models and Experimental Studies

Animal models, particularly genetically modified mice, allow researchers to study gene-environment interactions under controlled conditions. By exposing genetically predisposed animals to specific environmental factors, scientists can observe disease development and progression. These models are widely used in cancer, neurodegenerative disorders, and metabolic disease research (Chen et al., 2019).

Conclusion. The study of gene-environment interactions is vital for understanding hereditary genetic diseases. Methods such as epidemiological studies, twin analyses, GWAS, epigenetics, and animal models each contribute to uncovering the complex relationship between genetics and environmental factors. Future research integrating these approaches will enhance disease prediction, prevention, and personalized treatment strategies, ultimately improving public health outcomes.

References:

1. G., Jalilova Gulchekhra, N., Rasulova Nilufar, N., Mukhamedova Nigora, L., Kravchenko, Lenara. BIO Web of Conferences //Impact of environmental factors in current industrial development 138,03041
2. Boomsma, D. I., Busjahn, A., & Peltonen, L. (2002). Classical twin studies and beyond. *Nature Reviews Genetics*, 3(11), 872-882.
3. Brenner, H., Blettner, M., & Gefeller, O. (2017). Epidemiological methods in studying gene-environment interactions. *International Journal of Epidemiology*, 46(3), 749-758.
4. Chen, H., Uryu, K., & Bi, W. L. (2019). Animal models for studying genetic and environmental interactions in disease. *Current Topics in Developmental Biology*, 132, 67-95.



5. Feil, R., & Fraga, M. F. (2012). Epigenetics and the environment: Emerging patterns and implications. *Nature Reviews Genetics*, 13(2), 97-109.
6. Lander, E. S., Linton, L. M., & Birren, B. (2001). Initial sequencing and analysis of the human genome. *Nature*, 409(6822), 860-921.
7. Visscher, P. M., Wray, N. R., Zhang, Q., et al. (2017). 10 years of GWAS discovery: Biology, function, and translation. *American Journal of Human Genetics*, 101(1), 5-22.