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Genetic Bases of Hereditary Metabolism Diseases

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Abstract: The nature of genotype-environment interactions and their role in the formation of predisposition to pathogenetically independent nosological forms of multifactorial pathology was studied by assessing the joint influence of the studied molecular genetic markers and environmental factors of a toxic nature on the risk of occurrence of the diseases in question. The role of biotransformation enzyme gene polymorphism was studied xenobiotics in the formation of predisposition to the joint development of multifactorial pathology and model intergenic interactions that determine various compatibility.

Keywords: Hereditary metabolic diseases, genetics, medical progress, gene pool structure.

Hereditary metabolic diseases are a large group of monogenically inherited human diseases. The practical significance of their research is determined by purely medical aspects, i.e. the need to develop methods for diagnosis, treatment, and prevention of these diseases, which represent a serious problem for modern healthcare. On the other hand, hereditary metabolic diseases - typical "biochemical" mutations of the human genome - are a powerful tool for studying normal human metabolism, which is characterized by great complexity and a large number of "blank spots" on its map.

Being one of the most rapidly developing branches of modern medical biochemical genetics, research into hereditary metabolic diseases is particularly focused on solving important medical problems (reducing childhood morbidity and mortality) and the development of genetics, in particular human genetics (study of the structural organization of the genome, the molecular mechanism of mutations, biochemical polymorphism of populations, mapping genes on chromosomes, etc.).

As is known, the progress of medicine and healthcare depends on many components, including the success of fundamental sciences, among which genetics occupies a prominent and sometimes leading place.

The contribution of genetics to clinical medicine especially increased in the last quarter of the 20th century. Human genetics in the 21st century is not only the molecular and clinical levels, but also the population level, which remains no less significant .

In the 20th century, many new factors and conditions appeared that changed human heredity, which he, as a biological species, did not encounter during his long evolution. This is population migration and the expansion of marriage boundaries, family planning in healthy individuals and reproductive compensation in families burdened with hereditary pathology.

As is known, the genetic diversity of a population is largely determined by demographic processes, which are not constant and change over time. Significant changes in the demographic structure of populations can be observed in the event of sudden socio-economic transformations

of society. This applies to such indicators as the level of birth and death rates of the population, the intensity and direction of migration, marriage structure, etc.

The dynamics of the gene pool are dictated not by heredity, but by the social process, which is primary. The structure of the gene pool changes in response to changes in society. When assessing the state of the gene pools of human populations in the past and present, to predict their development for the future, it is necessary to analyze the main factors of evolution - the factors of gene pool dynamics.

It is generally accepted that such parameters of population structure as population size, degree of geographic and ethnic isolation, its subdivision , genetic drift, selection pressure, gene migration and others have a direct impact on the prevalence, accumulation and spectrum of monogenic hereditary diseases.

The need to further improve the provision of medical genetic care to the population is obvious.

WHO expert assessments have clearly shown that a relatively inexpensive medical genetic service is economically beneficial compared to the social and financial burden from diseases that this service helps to avoid. Family planning, subject to medical and genetic counseling, is becoming more advanced. The contribution of human genetics to the effectiveness of medical genetic counseling is realized through more accurate diagnosis of diseases or heterozygous conditions, as well as a wider choice of solutions for the family - limitation of childbearing, prenatal , preimplantation diagnostics, pre- and postnatal. Data on the prevalence of heterozygous carriage in various populations and ethnic groups are becoming important for the prevention of NB.

The program for the prevention of congenital and hereditary diseases should be based on data on the burden of hereditary pathology. Already today, certain achievements can be noted: 1) accurate, effective and universal methods for diagnosing hereditary diseases at any stage of ontogenesis have been developed; 2) the experimental and clinical foundations of gene therapy for hereditary and non-hereditary diseases have been laid; 3) the molecular basis of preventive medicine, etc., is being actively developed.

Knowledge of the epidemiology of hereditary diseases in specific populations is necessary not only to identify the etiological factors and mechanisms that influence the incidence of these diseases in the population, but also the characteristics of the pathogenesis and possible methods of treatment, to develop more effective methods of combating them.

The study of the patterns of spread of hereditary diseases in various ethnic, racial and territorial groups of the population is of current importance and is ultimately aimed at the preservation and development of the gene pool of human populations. Therefore, studying the burden of hereditary pathology with the study of its quantitative and qualitative characteristics is important for all regions. Another criterion for changing the genetic load is a change in the spectrum of hereditary pathology.

A comparative analysis of the diversity of nosological forms relative to previous studies showed that the spectrum of hereditary pathology in the populations of the region has become narrower, mainly due to autosomal recessive forms. The genetic diversity of populations within population groups of the same scale can be dramatically different, since it is not determined by the scale itself, but by historical, demographic, social, and economic conditions of development. Establishing connections between changes in the natural and social environment of a person with ongoing changes in the gene pool of the population requires determining the nature of the dynamics of the gene pool over time.

Multifactorial diseases (MFDs) represent the largest and most diverse group of diseases, accounting for more than 90% of all human somatopathology and characterized by the highest growth rates of morbidity, mortality and disability among the working population in modern populations. WHO researchers have shown that the implementation of treatment and preventive

measures is no longer able to change the emerging situation, and traditional approaches to the treatment of common multifactorial diseases lead to colossal economic costs and very modest results.

The problem of low effectiveness of treatment and preventive measures is associated with the lack of their etiological focus due to insufficient understanding of the key mechanisms of formation of the vast majority of multifactorial diseases.

Over the past decade, the global scientific community has made enormous efforts to study the etiology and pathogenesis of multifactorial pathology of various organs and systems of the body.

Numerous foreign and domestic studies have shown that complex interactions of genetic and environmental factors underlie the occurrence of MD. With the development of molecular genetic technologies, broad prospects have opened up for formalizing the genetic component of susceptibility to multifactorial diseases. Currently, a significant amount of data has been accumulated on the involvement of various polymorphic genes in the formation of predisposition to multifactorial pathology.

However, despite the achievements of the global scientific community in the field of studying the human genome and in the development of high-resolution methods for DNA analysis, a relatively small number of genes are still known, which together only partially explain individual links in the pathogenesis of some multifactorial diseases.

Often, in genetic studies of multifactorial diseases, environmental effects, which are of paramount importance specifically for this class of diseases, are leveled out or ignored. Proof of the importance of the environmental component is the rapid increase in recent years in the frequency of many multifactorial diseases in populations, which cannot be explained by changes in the genetic component in such a short period of time from an evolutionary point of view.

The accumulated numerous data on environmental risk factors for multifactorial diseases are often contradictory, do not fully explain the mechanisms of their pathological influence on the body and do not allow us to formulate a unified concept of the etiology of common diseases in humans. Currently, there is an increasing interest among researchers in studying the effects of chemical pollution of the environment, the level of which has been continuously increasing in economically developed countries since the middle of the last century, on the formation of common diseases. Today, more than 5 million chemical substances are already known (atmospheric pollutants, pesticides, pharmaceuticals, cosmetics, food additives, bad habits, etc.), to which humans are constantly exposed, and for many of which an etiological connection with MD is shown.

For environmental genetics, it is becoming increasingly important to identify in different populations specific genes and environmental factors, the interaction of which forms the norm of a person's resistance reaction and his adaptation to a changing environment. In this regard, the most suitable genetic markers for ecogenetic studies of MD are polymorphic variants of biotransformation enzyme genes xenobiotics (XBK), the expression of which, unlike other classes of genes, is directly regulated by the influence of environmental factors of a chemical nature. However, despite the ever-increasing environmental pollution, the high level of chemicalization of industry, agriculture and everyday life, the study of ecogenetic mechanisms of the formation of multifactorial diseases is still away from the main vector of scientific research. An analysis of numerous studies on the associations of various classes of genes with cancer susceptibility showed that more than 70% of consistently reproducible positive gene mapping results were obtained through the study of polymorphism of the FBK genes, thereby demonstrating the significant contribution of the ecogenetic component to the susceptibility to malignant neoplasms. At the same time, studies of polymorphism of FBK genes in common non-oncological diseases are few. Comprehensive assessment of the involvement of polymorphic variants of biotransformation enzyme genes xenobiotics in the formation of multifactorial pathology of non-oncological nature has not yet been carried out.

LITERATURES

1. Zhabborova, O. I., & Kenjaeva, H. P. (2018). Bases of gender equality of rights in Uzbekistan. *Международный журнал гуманитарных и естественных наук*, (5-2), 18-21.
2. Asatullayev, A. N., & Jabborova, O. G. (2022). Bleeding and its Types, Organization of Emergency Assistance in Bleeding. *European Journal of Life Safety and Stability (2660-9630)*, *13*, 111-116.
3. HUNTING, T., & REGION, A. (2020). RR Rakhmonov, PhD, Bukhara State Medical Institute, Bukhara OI Jabborova, PhD, Bukhara State Medical Institute, Bukhara MM Turawev, PhD, Bukhara State University, Bukhara. *ХОРАЗМ МАЪМУН АКАДЕМИЯСИ АХБОРОТНОМАСИ*, *9*.
4. Жабборова, О. И., & Кенжаева, Х. П. (2018). Экологические мировоззрения Ибн Сины. *Международный журнал гуманитарных и естественных наук*, (5-2), 22-24.
5. Zh, A. L., Abdullaeva, M. A., Sh, I. A., Jabborova, O. I., Nigmatullaeva, M. A., Kudratova, M. O., & Navruzova, U. O. (2021). Ways to solve the incidence of covid-19 as a global problem. *Annals of the Romanian Society for Cell Biology*, 1873-1880.
6. Asimov, I. (1975). *The biological sciences*. Penguin Books.
7. Nazarova, F. A., & Jabborova, O. (2021). Protection of Atmospheric Air and its Role in Nature. *Central Asian Journal of Theoretical and Applied Science*, *2*(10), 139-143.
8. Жабборова, О. И. (2018). Толерантность-как нравственное качество. *Наука, техника и образование*, (12 (53)), 92-94.
9. Jabborova, O. I., & Iskandarov, U. E. (2021). Study of the immunology of complications after kidney transplantation. *Central Asian Journal of Medical and Natural Science*, *2*(3), 99-101.
10. Жабборова, О. И., & Абдуллаева, М. А. (2023). КЛИМАКТЕРИК ДАВРДАГИ ОСТЕОПОРОЗНИ БАШОРАТ ҚИЛИШ БЕЛГИЛАРИ. *Биология*, (3), 144.
11. Iskandarovna, J. O., & Ulugbek, I. (2022). The Main Directions of Teaching" Medical Biology". *INTERNATIONAL JOURNAL OF INCLUSIVE AND SUSTAINABLE EDUCATION*, *1*(4), 256-260.
12. Жабборова, О. И. (2021). ВАЖНЫЕ НАПРАВЛЕНИЯ ИЗУЧЕНИЯ ВЛИЯНИЯ ХИМИЧЕСКИХ И ФИЗИЧЕСКИХ ФАКТОРОВ НА ИММУННУЮ СИСТЕМУ ОРГАНИЗМА. *Вестник науки и образования*, (2-3 (105)), 4-6.
13. Жабборова, О. И. (2023). ТАЪЛИМ ВА ТАРБИЯНИНГ МАЪНАВИЙ АСОСЛАРИ. *TA'LIM VA RIVOJLANISH TAHLILI ONLAYN ILMIY JURNALI*, *3*(9), 181-187.
14. Жабборова, О. И. (2023). Профессор-Ўқитувчилар Таркибининг Учякдиллигини Ривожлантиришда Академик Тиббиёт Марказлари Ва Университет Клиникаларининг Ўрни. *AMALIY VA TIBBIYOT FANLARI ILMIY JURNALI*, *2*(1), 31-36.
15. Iskandarovna, J. O., & Ulugbek, I. (2022). Interactive Learning Methods in Teaching Biology. *Web of Synergy: International Interdisciplinary Research Journal*, *1*(2), 11-17.
16. Iskandarovna, J. O., & Abdullaevich, N. N. (2022). ЎТКИР НУРЛАНИШНИНГ БАКТЕРИАЛ ТРАНСЛОКАЦИЯ ФЕНОМЕНИГА ТАЪСИРИ ХУСУСИЯТЛАРИ ТАВСИФИ. *JOURNAL OF BIOMEDICINE AND PRACTICE*, *7*(5).
17. Нуралиев, Н. А., & Жабборова, О. И. (2022). Анализ Результатов Изучения Влияния Острого Облучения На Нормальную Микрофлору Толстого Кишечника Лабораторных

Животных В Динамике. *Central Asian Journal of Medical and Natural Science*, *3*(5), 396-403.

18. Iskandarovna, J. O., Husniddinovna, S. S., & Raximovich, R. R. (2022). BUXORO VILOYATI SHAROITIDA XONQIZI (COCCINELIDAE) NING TARQALISHI, BIOLOGIYASI VA TABIATDAGI AHAMIYATI. *BARQARORLIK VA YETAKCHI TADQIQOTLAR ONLAYN ILMIY JURNALI*, 721-727.
19. Нуралиев, Н. А., & Жабборова, О. И. (2022). ПОКАЗАТЕЛИ ВЫСЕВАЕМОСТИ МИКРООРГАНИЗМОВ, ТРАНСЛОЦИРОВАВШИЕСЯ ИЗ ТОЛСТОГО КИШЕЧНИКА ВО ВНУТРЕННИЕ ОРГАНЫ ПОД ВЛИЯНИЕМ ОСТРОГО ОБЛУЧЕНИЯ В ЭКСПЕРИМЕНТЕ. In *Актуальные вопросы экспериментальной микробиологии: теория, методология, практика, инноватика* (pp. 157-160).
20. Жабборова, О. И. (2022). БУХОРО ВИЛОЯТИ КОКЦИНЕЛЛИДЛАРИНИНГ БИОЕКОЛОГИЯСИ. *Scientific progress*, *3*(2), 536-539.
21. Жабборова, О. И., & Худайкулова, Н. И. (2021). КОКЦИНЕЛЛИДЛАРНИНГ ИЛМИЙ АДАБИЁТЛАРДАГИ ТАҲЛИЛИ. *Журнал Биологии и Экологии*, *3*(1).
22. Jabborova, O. I. (2021). O ‘RTA ASR SHARQ MUTAFAKKIRLARI QARASHLARINING YOSHLAR AXLOQIY TARBIYASIDAGI O ‘RNI. *Oriental renaissance: Innovative, educational, natural and social sciences*, *1*(8), 26-31.
23. Жабборова, О. И. (2021). ЯНГИ ЎЗБЕКИСТОН: ДЕМОКРАТИЯ ВА ИНСОН ҲУҚУҚЛАРИ. *Scientific progress*, *2*(4), 789-796.
24. Жабборова, О. И. (2021). БУХОРО ВОҲАСИ ХОНҚИЗИ ҚЎНҒИЗЛАРИ. *Academic research in educational sciences*, *2*(3), 365-371.
25. Жабборова, О. И. (2020). РЕПАРАЦИЯ ДНК-ЖИЗНЕННО НЕОБХОДИМЫЙ И СЛОЖНО РЕГУЛИРУЕМЫЙ ПРОЦЕСС. *Наука и мир*, *2*(4), 8-10.
26. Жабборова, О. И. БУХОРО ВОҲАСИНИНГ ТАБИИЙ ШАРОИТИ ХУСУСИДА. *УЧЕНЫЙ XXI ВЕКА*, 8.
27. Жабборова, О. И. (2023). ШАРҚ МУТАФАККИРЛАРИ ТАБИБЛИК АХЛОҚИ ХУСУСИДА. *SIYOSATSHUNOSLIK, HUQUQ VA XALQARO MUNOSABATLAR JURNALI*, *2*(10), 45-51.
28. Жабборова, О. И. (2023). КОКЦИНЕЛЛИДЛАРНИНГ ЭКОЛОГИК ХУСУСИЯТЛАРИ ТАҲЛИЛИ. *AGROBIOTEXNOLOGIYA VA VETERINARIYA TIBBIYOTI ILMIY JURNALI*, *2*(9), 14-22.
29. Жабборова, О. И. (2023). Қишлоқ Хўжалиги Зараркунандаларига Қарши Биологик Курашнинг Асосий Жиҳатлари. *AMALIY VA TIBBIYOT FANLARI ILMIY JURNALI*, *2*(10), 64-70.
30. Жабборова, О. И. (2023). ЁШЛАР ТАРБИЯСИ ВА МАЪНАВИЙ МЕРОС. *TA'LIM VA RIVOJLANISH TAHLILI ONLAYN ILMIY JURNALI*, *3*(10), 182-189.
31. Жабборова, О. И. (2023). ОИЛА ИНСТИТУТИНИ МУСТАҲКАМЛАШНИНГ АСОСИЙ ОМИЛЛАРИ. *ARXITEKTURA, MUHANDISLIK VA ZAMONAVIY TEXNOLOGIYALAR JURNALI*, *2*(10), 18-23.
32. Жабборова, О. И. (2023). МИЛЛИЙ МАЪНАВИЙ МЕРОСДА ТАБИБЛИК АХЛОҚИ. *IJTIMOIY FANLARDA INNOVASIYA ONLAYN ILMIY JURNALI*, *3*(10), 42-51.
33. Жабборова, О. И. (2023). ТАЪЛИМ ТИЗИМИНИ ТУБДАН ТАКОМИЛЛАШТИРИШ БЎЙИЧА АМАЛГА ОШИРИЛГАН ЧОРА ТАДБИРЛАРНИНГ ЖАМОАТЧИЛИК ФИКРИНИ РИВОЖЛАНТИРИШДАГИ АҲАМИЯТИ. *BARQARORLIK VA YETAKCHI TADQIQOTLAR ONLAYN ILMIY JURNALI*, *3*(10), 99-106.

34. Iskandarovna, J. O. (2023). Scientific and Methodological Foundations of Teaching Personnel Training in the Higher Education System. *Best Journal of Innovation in Science, Research and Development*, *2*(10), 43-50.

35. Жабборова, О. И. (2023). ЯНГИ ЎЗБЕКИСТОН ШАРОИТИДА ИНСОН ҲУҚУҚЛАРИНИ ҲИМОЯ ҚИЛИШНИНГ АСОСИЙ МЕХАНИЗМЛАРИ. *BARQARORLIK VA YETAKCHI TADQIQOTLAR ONLAYN ILMIY JURNALI*, *3*(10), 25-32.

36. Iskandarovna, J. O. (2023). Scientific and Methodological Basis for Training Pedagogical Staff in the System of Higher Education. *Web of Synergy: International Interdisciplinary Research Journal*, *2*(1), 103-110.

37. Imomjonovich, I. I., & Amirkulovna, A. G. (2021). Methods of early detection of rejection in a kidney transplant from a relative donor. *Academicia Globe*, *2*(05), 293-295.

38. Imomjonovich, I. I., & Amirkulovna, A. G. (2021). Current immunological problems in kidney transplantation.

39. Arashova, G. A., & Kh, G. M. (2021). Morphological and Functional Aspects of the Lymphatic System and its Current Problems. *International Journal of Innovative Analyses and Emerging Technology*, *10*(5), 149-152.

40. Арашова, Г. А., & Бобожоджаев, С. Н. (2008). Клинико-эпидемиологические особенности бруцеллёза. In *Российская научно-практическая конференция* (No. 4 Часть 1, pp. 14-15).

41. Арашова, Г. А. (2019). К вопросу о течении ветряной оспы у взрослых. *Проблемы биологии и медицины*, (4-2), 10-12.

42. Арашова, Г. А. (2019). Клинические особенности течения ветряной оспы у взрослых. *Новый день в медицине*, (4), 130-132.

43. Arashova G.A. (2023). Clinical and Epidemiological Features of Acute Brucellosis at the Present Stage. *American Journal of Pediatric Medicine and Health Sciences (2993-2149)*, *1*(9), 80–84.

44. Gulnora Amirqulovna Arashova. (2023). PRESENT STAGE CLINICAL MANIFESTATIONS OF CHICKENPOX. *International Journal of Medical Sciences And Clinical Research*, *3*(08), 64–71.

45. Amirqulovna, A. G. (2023). CLINICAL FEATURES OF THE COURSE OF CHICKENPOX AT THE PRESENT STAGE. *INTERNATIONAL JOURNAL OF HEALTH SYSTEMS AND MEDICAL SCIENCES*, *2*(6), 74-79.

46. Amirkulovna, A. G. (2022). Observation of the Immuno-Pathogenetic State of Measles in Adults. *INTERNATIONAL JOURNAL OF HEALTH SYSTEMS AND MEDICAL SCIENCES*, *1*(5), 154-158.

47. Amirkulovna, A. G. (2022). Clinical and Epidemiological Features of Measles in Adults. *Central Asian Journal of Medical and Natural Science*, *3*(3), 80-86.

48. Шарипова, И. П., Эгамова, И. Н., Рахимова, В. Ш., & Арашева, Г. А. (1999). ПОКАЗАТЕЛИ ТРАНЗИЕНТНОЙ ЭЛАСТОГРАФИИ У БОЛЬНЫХ С ПОРАЖЕНИЕМ ПЕЧЕНИ ВГС ЭТИОЛОГИИ ПОСЛЕ ПРОТИВОВИРУСНОЙ ТЕРАПИИ. *ИНФЕКЦИЯ, ИММУНИТЕТ и ФАРМАКОЛОГИЯ*, 298.

49. Arashova Gulnora Amirkulovna. (2023). THE ROLE OF MEDICAL CULTURE IN THE PREVENTION OF INFECTIOUS DISEASES. *Best Journal of Innovation in Science, Research and Development*, 605–610.

50. Arashova Gulnora Amirkulovna. (2023). Modern Methods of Combating Infectious Diseases. *American Journal of Pediatric Medicine and Health Sciences (2993-2149)*, *1*(10), 70–75.
51. https://www.dissercat.com/content/dinamika-raspredelennosti-nasledstvennoi-patologii-v-tomskoi-obl
52. https://www.dissercat.com/content/polimorfizm-genov-fermentov-biotransformatsii-ksenobiotikov-i-ikh-kompleksnoe-vliyanie-na-pr