# GEN KASALLIKLARI: AMINOKISLOTALAR ALMASHINUVINING BUZILISHI BILAN BOG‘LIQ BO‘LGAN KASALLIKLAR

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***Annotatsiya.*** *Ushbu maqolada aminokislotalar almashinuvining buzilishi bilan bog‘liq genetik kasalliklar tizimli tarzda tahlil qilinadi. Aminokislotalarning normal metabolizmi organizmning hayotiy jarayonlari uchun muhim ahamiyatga ega bo‘lib, uning buzilishi turli klinik simptomlar va surunkali kasalliklarni keltirib chiqaradi. Fenilketonuriya, homosistinuriya, tirozinemiya va boshqa metabolik buzilishlar nevrologik buzilishlar, jigar va buyrak disfunktsiyalari hamda kardiometabolik muammolar bilan bog‘liq ekanligi aniqlangan.*

*Maqolada ushbu kasalliklarning patofiziologiyasi, molekulyar mexanizmlari, genetik asoslari va diagnostika usullari batafsil ko‘rib chiqiladi. Shu bilan birga, genetik testlar va metabolik monitoring orqali kasalliklarni erta aniqlash, individual davolash strategiyalarini ishlab chiqish imkoniyatlari muhokama qilinadi.*

***Kalit so‘zlar:*** *genetik kasalliklar, aminokislota almashinuvi, fenilketonuriya, homosistinuriya, tirozinemiya, metabolik buzilishlar, molekulyar diagnostika.*

# ГЕНЕТИЧЕСКИЕ ЗАБОЛЕВАНИЯ: ЗАБОЛЕВАНИЯ, СВЯЗАННЫЕ С НАРУШЕНИЕМ ОБМЕНА АМИНОКИСЛОТ

***Аннотация.*** *В статье систематически рассматриваются генетические заболевания, связанные с нарушением обмена аминокислот. Нормальный метаболизм аминокислот имеет ключевое значение для жизненно важных процессов организма, и его нарушение приводит к развитию различных клинических симптомов и хронических заболеваний. Фенилкетонурия, гомоцистинурия, тирозинемия и другие метаболические расстройства связаны с неврологическими нарушениями, дисфункцией печени и почек, а также кардиометаболическими проблемами. Статья подробно рассматривает патофизиологию, молекулярные механизмы, генетическую основу и методы диагностики этих заболеваний. Также обсуждаются возможности раннего выявления и индивидуализированного лечения с помощью генетического тестирования и метаболического мониторинга.*

***Ключевые слова:*** *генетические заболевания, обмен аминокислот, фенилкетонурия, гомоцистинурия, тирозинемия, метаболические нарушения, молекулярная диагностика.*

# GENETIC DISORDERS: DISEASES ASSOCIATED WITH AMINO ACID METABOLISM DISRUPTION

***Article.*** *This article provides a systematic review of genetic disorders associated with amino acid metabolism dysfunction. Normal amino acid metabolism is essential for vital physiological processes, and its disruption can lead to various clinical symptoms and chronic diseases.*

*Disorders such as phenylketonuria, homocystinuria, tyrosinemia, and other metabolic abnormalities are associated with neurological impairments, hepatic and renal dysfunction, and cardiometabolic complications. The article details the pathophysiology, molecular mechanisms, genetic basis, and diagnostic approaches of these disorders. Additionally, it discusses early detection strategies and individualized treatment options through genetic testing and metabolic monitoring.*

***Keywords:*** *genetic disorders, amino acid metabolism, phenylketonuria, homocystinuria, tyrosinemia, metabolic disorders, molecular diagnostics.*

**Introduction**

Amino acids are molecules that participate in the body's primary metabolic processes, and their normal metabolism is essential for maintaining vital physiological functions [1]. Studies have shown that disruptions in amino acid metabolism lead to various clinical manifestations, including neurological, hepatic, and renal dysfunctions [2]. Genetic disorders such as phenylketonuria, homocystinuria, and tyrosinemia arise as a result of impaired amino acid metabolism, and their pathophysiology is explained through various molecular mechanisms [3].

Furthermore, metabolic disturbances negatively affect not only physical health but also psychological and cognitive functions, thereby reducing patients' quality of life [4]. Research indicates that early detection of these disorders and the implementation of metabolic monitoring are effective in preventing cognitive and physical complications [5]. In addition, genetic testing and molecular diagnostic methods play a crucial role in disease identification and in the development of individualized treatment strategies [6].

This article systematically examines the molecular mechanisms, clinical manifestations, and diagnostic approaches of genetic disorders associated with amino acid metabolism disruption.

Moreover, contemporary studies emphasize the potential of metabolic and genetic monitoring to improve patient prognoses [2]. Research demonstrates that a thorough investigation of the relationship between genetic disorders and metabolic disturbances aids in the early detection of diseases and the optimization of therapeutic strategies [1].

In addition, global statistical data on these disorders highlight the importance of genetic and metabolic monitoring in patients, which plays a significant role in health promotion and the development of preventive measures [3]. Therefore, studying genetic disorders associated with impaired amino acid metabolism is a pressing issue in modern medicine and genetic research [5].

In conclusion, this article aims to provide a systematic study of genetic disorders linked to amino acid metabolism disruption and to establish a scientific foundation for their diagnosis, treatment strategies, and preventive interventions [6].

**Materials and methods**

The study employed a mixed-methods approach to investigate genetic disorders associated with disruptions in amino acid metabolism. Primary data were collected through genetic testing, metabolic analyses, clinical observations, and literature review. During the study, the molecular mechanisms, clinical manifestations, and diagnostic possibilities of disorders related to impaired amino acid metabolism, including phenylketonuria, homocystinuria, and tyrosinemia, were evaluated.

Both laboratory and observational methods were used in combination during the analysis.

In the laboratory phase, participants underwent genetic testing and biochemical analyses, with monitoring of amino acid metabolism and enzymatic activity. During the observational phase, patients' clinical presentations, symptoms, and cognitive functions were assessed. In addition, the study examined the potential for early disease detection and the development of individualized treatment strategies.

The data were analyzed using both qualitative and quantitative methods. Results from genetic and metabolic monitoring were employed to evaluate patient prognosis and optimize management of metabolic disturbances. The combination of clinical and laboratory observations facilitated a systematic study of genetic disorders associated with disruptions in amino acid metabolism.

All stages of the research were focused on identifying genetic bases, enabling early diagnosis, implementing metabolic monitoring, and developing individualized treatment strategies. This methodology provides a framework for systematically studying disorders associated with amino acid metabolism disruption in modern medicine and molecular genetic research.

**Results and discussion**

The study results indicate that genetic disorders associated with disruptions in amino acid metabolism are linked to neurological, hepatic, and renal dysfunctions, as well as cognitive impairments and psychological symptoms. In patients with phenylketonuria, neurological disturbances and slowed intellectual development were observed, whereas homocystinuria was associated with cardiovascular and skeletal system complications. Tyrosinemia was characterized by impaired liver function and various metabolic complications.

Findings from metabolic monitoring and genetic testing demonstrated that early disease detection and the development of individualized treatment strategies are feasible. These approaches were shown to be effective in improving patients' cognitive and physical functions, reducing symptoms, and preventing chronic complications.

Furthermore, the results highlighted that the molecular mechanisms underlying disruptions in amino acid metabolism are closely associated with the clinical manifestations of these disorders.

Enzymatic deficiencies and genetic mutations determine the severity of the disease and the spectrum of symptoms, which is crucial for designing individualized treatment and preventive strategies.

Analysis also revealed that disturbances in amino acid metabolism not only affect individual health but also negatively impact patients' social and academic activities. Therefore, metabolic and genetic monitoring, along with personalized treatment plans, play a central role in disease management.

Overall, the study results underscore the importance of systematically investigating genetic disorders associated with amino acid metabolism disruption, providing a scientific foundation for their diagnosis, treatment strategies, and preventive interventions.

**Conclusion**

The study demonstrates that genetic disorders associated with disruptions in amino acid metabolism have significant adverse effects on the neurological, hepatic, renal, and cardiovascular systems. Disorders such as phenylketonuria, homocystinuria, and tyrosinemia cause clinical symptoms and chronic complications through metabolic disturbances.

Metabolic monitoring, genetic testing, and individualized treatment strategies have been shown to be effective in early disease detection and symptom reduction. Therefore, the study of genetic disorders linked to impaired amino acid metabolism remains a pressing issue in modern medicine and molecular genetic research.

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