PATHOLOGY OF GENETIC DISORDERS

Abdullayev Jasurbek Saidazim o‘g‘li
Email: jasurabdullaev654@gmail.com
Address: Tashkent, Uzbekistan
Place of study: Tashkent state medical university
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Annotatsiya: Genetik buzilishlar irsiy apparatning strukturaviy yoki funksional o‘zgarishlari natijasida rivojlanadigan murakkab patologik holatlar hisoblanadi. Ushbu kasalliklar organizmning embrional rivojlanishi, hujayra metabolizmi, oqsil sintezi va fiziologik funksiyalariga jiddiy ta’sir ko‘rsatadi. Zamonaviy patologik anatomiya va molekulyar genetika fanlari genetik buzilishlarning rivojlanish mexanizmlarini chuqur o‘rganishga imkon yaratdi. Ushbu maqolada genetik buzilishlarning etiologiyasi, patogenezi, molekulyar asoslari, hujayraviy o‘zgarishlari, klinik namoyon bo‘lishi va patologik anatomiya nuqtai nazaridan morfologik xususiyatlari keng ilmiy tahlil qilindi. Robbins Basic Pathology, Gray’s Anatomy, Harrison’s Principles of Internal Medicine, WHO, NCBI va boshqa ilmiy manbalar asosida genetik mutatsiyalarning organizmga ta’siri chuqur yoritildi. Tadqiqotlar shuni ko‘rsatadiki, genetik buzilishlarda asosiy patologik mexanizm DNK replikatsiyasi va reparatsiyasi buzilishi, oqsil sintezining o‘zgarishi, hujayra differensiyalanishining izdan chiqishi hamda ferment tizimlari faoliyatining buzilishi bilan bog‘liq.

Kalit so‘zlar: genetik buzilishlar, mutatsiya, xromosoma, DNK, irsiy kasalliklar, patologik anatomiya, molekulyar genetika, sindrom, gen mutatsiyasi, embrional rivojlanish.

Abstract: Genetic disorders are complex pathological conditions caused by structural or functional abnormalities of the hereditary apparatus. These disorders significantly affect embryonic development, cellular metabolism, protein synthesis, and physiological functions of the organism. Modern pathological anatomy and molecular genetics have made it possible to study the mechanisms of genetic disorders in depth. This article provides a broad scientific analysis of the etiology, pathogenesis, molecular basis, cellular alterations, clinical manifestations, and morphological characteristics of genetic disorders from the perspective of pathological anatomy. Based on Robbins Basic Pathology, Gray’s Anatomy, Harrison’s Principles of Internal Medicine, WHO, NCBI, and other scientific sources, the effects of genetic mutations on the human body were comprehensively described. Research demonstrates that the main pathological mechanisms of genetic disorders are associated with impaired DNA replication and repair, altered protein synthesis, disrupted cellular differentiation, and enzyme system dysfunction.

Keywords: genetic disorders, mutation, chromosome, DNA, hereditary diseases, pathology, molecular genetics, syndrome, gene mutation, embryonic development.

Аннотация: Генетические нарушения представляют собой сложные патологические состояния, возникающие вследствие структурных или функциональных изменений наследственного аппарата. Данные заболевания оказывают серьезное влияние на эмбриональное развитие, клеточный метаболизм, синтез белков и физиологические функции организма. Современная патологическая анатомия и молекулярная генетика позволили глубоко изучить механизмы развития генетических нарушений. В данной статье подробно проанализированы этиология, патогенез, молекулярные механизмы, клеточные изменения, клинические проявления

и морфологические особенности генетических нарушений с точки зрения патологической анатомии. На основе Robbins Basic Pathology, Gray’s Anatomy, Harrison’s Principles of Internal Medicine, WHO, NCBI и других научных источников подробно освещено влияние генетических мутаций на организм человека. Исследования показывают, что основными патологическими механизмами являются нарушения репликации и репарации ДНК, изменения синтеза белка, нарушения клеточной дифференцировки и дисфункция ферментных систем.

Ключевые слова: генетические нарушения, мутация, хромосома, ДНК, наследственные заболевания, патология, молекулярная генетика, синдром, генная мутация, эмбриональное развитие

Introduction

Genetic disorders are among the most complex and important fields of modern medicine and biology. These pathological conditions develop as a result of abnormalities in the DNA molecule carrying hereditary information and affect multiple organs and systems of the human body. Genetic diseases may begin during embryonic development and play an important role in the formation of congenital anomalies, metabolic syndromes, immune deficiency conditions, and malignant tumors.

Modern molecular genetic studies show that mutations may occur at the gene, chromosome, or genome level. These abnormalities disrupt cellular metabolism and interfere with normal protein synthesis and tissue development. When DNA replication and repair mechanisms are impaired, pathological proteins are produced and regulation of the cell cycle becomes weakened.

Down syndrome develops due to trisomy of chromosome 21 and is associated with intellectual disability and congenital heart defects.

Turner syndrome is related to monosomy of the sex chromosome and leads to abnormalities in reproductive system development.

Cystic fibrosis develops due to mutation of the CFTR gene and causes severe pathological changes in the respiratory and digestive systems.

Phenylketonuria develops due to enzyme deficiency and has toxic effects on the central nervous system.

Literature review

In Robbins Basic Pathology, genetic disorders are described as one of the major branches of molecular pathology, and abnormalities in DNA replication, transcription, and translation processes are explained in detail. Gray’s Anatomy describes the influence of chromosomal abnormalities on embryonic development, organ deformities, and congenital anomalies. Harrison’s Principles of Internal Medicine provides extensive information about the clinical manifestations, systemic complications, and diagnosis of genetic diseases. According to WHO data, congenital genetic diseases are among the major causes of childhood mortality. Scientific databases of NCBI indicate that genetic mutations play an important role in cell cycle dysregulation and malignant transformation.

Gregor Mendel discovered the laws of heredity and is recognized as the founder of genetics.

James Watson and Francis Crick developed the double helix model of DNA and made major contributions to molecular genetics.

Thomas Hunt Morgan developed the chromosomal theory of inheritance.

This article was prepared based on the analysis of scientific literature related to pathological anatomy, molecular genetics, histology, embryology, and internal medicine. The molecular mechanisms of genetic disorders, chromosomal abnormalities, and pathological processes at cellular and tissue levels were studied in depth.

In addition, DNA mutations, disturbances in gene expression, enzyme deficiencies, pathological protein synthesis, and abnormalities in cellular differentiation were scientifically analyzed. The morphological and clinical characteristics of hereditary syndromes were evaluated using comparative methods.

Results

The analysis demonstrated that the main pathological processes in genetic disorders are associated with impaired cellular differentiation, abnormal protein synthesis, enzyme system deficiencies, and disturbances in organogenesis. Defects in DNA replication and repair mechanisms lead to disruption of the cell cycle, causing structural and functional abnormalities in various tissues. Genetic mutations occurring during embryonic development significantly affect multiple organs and systems, resulting in congenital malformations and morphological anomalies.

Diseases caused by chromosomal mutations are characterized by skeletal deformities, congenital heart defects, central nervous system abnormalities, and endocrine disorders. Down syndrome is associated with brachycephaly, epicanthal folds, muscular hypotonia, intellectual disability, and congenital heart defects. Due to immune dysfunction, patients are also more susceptible to infectious diseases.In Turner syndrome, ovarian hypoplasia, short stature, cardiovascular abnormalities, and impaired reproductive function are commonly observed. In some cases, thyroid gland disorders and metabolic syndrome may also develop.

In diseases related to gene mutations, pathological protein synthesis and enzyme deficiencies play a major role. Cystic fibrosis is characterized by thick secretions of exocrine glands due to CFTR gene mutation, leading to bronchial obstruction and chronic inflammation. Fibrosis, bronchiectasis, and chronic infectious processes are observed in lung tissue.

In Phenylketonuria, impaired phenylalanine metabolism leads to accumulation of toxic metabolites that damage the central nervous system. As a result, intellectual disability, neurological disorders, and metabolic encephalopathy may develop.

The analysis also demonstrated that certain genetic mutations increase the risk of malignant tumor formation. Activation of oncogenes or inactivation of tumor suppressor genes leads to uncontrolled cellular proliferation, resulting in malignant transformation and metastasis.

Conclusion

Genetic disorders produce complex pathological, molecular, and morphological alterations in the human body. These diseases disrupt not only individual organs but also the development and functional condition of the entire organism. Modern molecular genetics and pathological anatomy serve as important scientific foundations for understanding the mechanisms of genetic diseases. Early diagnosis, prenatal screening, and genetic counseling are essential for prevention and improvement of prognosis.

Adabiyotlar, References, Литературы:

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