CONGENITAL ANOMALIES AND DISORDERS IN PREMATURE NEWBORNS

Author: ***Turaqulova Zabarjat***
Email:zabarjatturaqulova004@gmail.com
Address: Tashkent, Uzbekistan
Educational institution: Tashkent state medical university

Annotatsiya: Tug‘ma anomaliyalar va chala tug‘ilish (prematur tug‘ilish) homila rivojlanishining intrauterin davrida genetik, xromosoma, epigenetik va teratogen omillar ta’sirida yuzaga keladigan murakkab patologik holatlardir. Ushbu maqolada embriogenez buzilishlari, organogenez nuqsonlari hamda prematur chaqaloqlarda ko‘p tizimli yetilmaganlik holatlari chuqur tahlil qilindi. Nafas olish, yurak-qon tomir, nerv va immun tizimlarining yetilmaganligi patologik anatomiya va neonatologiya nuqtai nazaridan yoritildi.

Kalit so‘zlar: tug‘ma anomaliya, prematur chaqaloq, teratogen, embriogenez, neonatal patologiya

Аннотатция: Врожденные аномалии и недоношенность являются сложными патологическими состояниями, возникающими во внутриутробном периоде развития плода под влиянием генетических, хромосомных, эпигенетических и тератогенных факторов. В статье подробно рассмотрены нарушения эмбриогенеза, дефекты органогенеза и мультисистемная незрелость у недоношенных новорожденных. Особое внимание уделено недостаточности дыхательной, сердечно-сосудистой, нервной и иммунной систем с позиции патологической анатомии и неонатологии.

Ключевые слова: врожденные аномалии, недоношенность, тератогены, эмбриогенез, неонатальная патология

Annotarion: Congenital anomalies and prematurity are complex pathological conditions arising during intrauterine fetal development due to genetic, chromosomal, epigenetic, and teratogenic factors. This article provides a detailed analysis of embryogenesis disorders, organogenesis defects, and multisystem immaturity in premature newborns. Special attention is given to respiratory, cardiovascular, nervous, and immune system insufficiency from the perspective of pathological anatomy and neonatology.

Keywords: congenital anomalies, prematurity, teratogens, embryogenesis, neonatal pathology

Introduction

Congenital anomalies and prematurity represent one of the most important and complex problems in modern medicine, particularly in the fields of pathology, embryology, and neonatology. These conditions arise during the earliest stages of human development when organogenesis and tissue differentiation are highly sensitive to both genetic and environmental influences. Even minimal disturbances in cellular proliferation, migration, or programmed cell death can lead to severe structural malformations or functional impairments in the developing fetus.

Prematurity, defined as birth before 37 completed weeks of gestation, is associated with incomplete maturation of multiple organ systems. The degree of immaturity directly correlates with the severity of clinical complications observed in newborns. The most critically affected systems include the respiratory, cardiovascular, central nervous, and immune systems. Among these, respiratory

insufficiency remains the leading cause of neonatal morbidity and mortality due to underdevelopment of alveolar structures and surfactant deficiency.

Respiratory distress syndrome of newborn is one of the most frequent and life-threatening complications in premature infants, primarily caused by insufficient surfactant production and immature lung architecture. This condition leads to impaired gas exchange, hypoxemia, and progressive respiratory failure if not managed promptly.

Literature review

According to standard pathological and embryological sources such as Robbins Basic Pathology and Langman's Medical Embryology, congenital anomalies originate from disruptions in tightly regulated developmental processes. These include genetic mutations, chromosomal abnormalities, and exposure to teratogenic factors during critical periods of embryogenesis, particularly between the third and eighth weeks of gestation.

Normal embryonic development requires precise coordination of gene expression, cellular signaling pathways, and tissue interactions. Disruption of these processes results in structural malformations involving major organ systems such as the heart, brain, kidneys, and skeletal structures. Genetic causes include aneuploidies, deletions, duplications, and point mutations affecting essential developmental genes.

Epigenetic mechanisms also play a significant role in congenital disorders. Alterations in DNA methylation and histone modification can lead to abnormal gene expression without changes in the DNA sequence itself. These mechanisms are increasingly recognized as important contributors to developmental abnormalities.

Teratogenic influences such as alcohol exposure, certain medications, viral infections, and ionizing radiation can interfere with cell proliferation and

differentiation. These agents are particularly harmful during periods of rapid organ formation, leading to irreversible developmental defects.

Victor A. McKusick is widely recognized for his foundational work in medical genetics, contributing significantly to the classification and understanding of hereditary and congenital disorders.

According to WHO data, congenital anomalies remain a leading cause of infant mortality and long-term disability worldwide, especially in low- and middle-income countries where prenatal screening and early intervention are limited.

Materials and methods

This study is based on a comprehensive review of scientific literature in embryology, pathological anatomy, medical genetics, and neonatology. The analysis focused on understanding the multilevel mechanisms involved in congenital anomalies and prematurity, including molecular, cellular, tissue, and organ system levels.

The following pathological processes were evaluated in detail:

1.Stages of embryogenesis and organogenesis

2.Genetic and chromosomal abnormalities affecting fetal development

3.Epigenetic regulation and gene expression alterations

4.Effects of teratogenic agents on developing tissues

5.Functional immaturity of organ systems in premature newborns

6.Neonatal adaptation mechanisms and their failure

Histopathological aspects such as cellular immaturity, vascular fragility, metabolic instability, and impaired immune response were also analyzed to understand structural-functional correlations in affected newborns.

Results

The analysis demonstrates that congenital anomalies and prematurity are multifactorial conditions involving complex interactions between genetic, epigenetic, and environmental factors.

At the genetic level, mutations in critical developmental genes and chromosomal abnormalities disrupt normal embryonic patterning and organ formation. These alterations result in structural malformations affecting the cardiovascular system, central nervous system, skeletal structures, and internal organs.

Epigenetic dysregulation further contributes to abnormal development by altering gene expression during key stages of organogenesis. These changes may persist throughout fetal development and after birth.

In premature newborns, the most significant pathological findings include:

1.Immature lung development leading to inadequate gas exchange

2.Cardiovascular instability with poor circulatory adaptation

3.Central nervous system immaturity with risk of intracranial hemorrhage

4.Underdeveloped immune system resulting in increased susceptibility to infections

5.Hepatic and metabolic enzyme immaturity affecting detoxification processes

Intraventricular hemorrhage of newborn is a severe complication frequently observed in premature infants due to fragile cerebral vasculature and unstable hemodynamic conditions.

From a pathological anatomy perspective, affected tissues show delayed maturation, fragile vascular structures, reduced cellular differentiation, and insufficient functional adaptation to extrauterine life.

Discussion

The findings highlight that congenital anomalies are primarily the result of disrupted developmental programming during embryogenesis. The severity of anomalies depends on the timing, duration, and intensity of the damaging factors. Early embryonic exposure typically results in more severe structural defects compared to later exposure during fetal development.

Prematurity significantly increases the risk of neonatal complications due to incomplete organ maturation. The lungs, in particular, are highly vulnerable due to insufficient surfactant production, leading to respiratory distress and hypoxia. Similarly, immaturity of the brain increases the risk of neurological damage, including hemorrhagic and ischemic lesions.

The interaction between genetic predisposition and environmental factors plays a crucial role in determining the final clinical outcome. Early detection, prenatal screening, and improved neonatal intensive care significantly improve survival rates and reduce long-term complications.

Conclusion

Congenital anomalies and prematurity represent complex medical conditions arising from disruptions in early human development. Their etiology involves

genetic, chromosomal, epigenetic, and environmental factors that interfere with normal embryogenesis and organogenesis.

Premature newborns are particularly vulnerable due to the immaturity of vital organ systems, leading to life-threatening complications. Early diagnosis, preventive strategies, and advanced neonatal care are essential for improving outcomes and reducing mortality.

A deeper understanding of the pathological mechanisms underlying these conditions is crucial for developing effective preventive and therapeutic approaches in modern medicine.

References

1. Robbins and Cotran Pathologic Basis of Disease. Kumar V, Abbas AK, Aster JC. Elsevier.

https://www.elsevier.com

2. Robbins Basic Pathology. Kumar V, Abbas AK, Aster JC. Elsevier. https://www.elsevier.com

3. Langman's Medical Embryology. Sadler TW. Wolters Kluwer. https://shop.lww.com

4. Nelson Textbook of Pediatrics. Kliegman RM et al. Elsevier. https://www.elsevier.com

5. Harrison's Principles of Internal Medicine. Jameson JL et al. McGraw-Hill. https://accessmedicine.mhmedical.com

6. World Health Organization (WHO). Congenital anomalies factsheet. https://www.who.int

7. Centers for Disease Control and Prevention (CDC). Birth defects information. https://www.cdc.gov

8. International Agency for Research on Cancer (IARC). Global health data. https://www.iarc.who.int

9. McKusick VA. Medical Genetics and OMIM database.

https://omim.org

10. Moore KL, Persaud TVN. The Developing Human: Clinically Oriented Embryology.

https://www.elsevier.com

11. Junqueira LC. Basic Histology: Text and Atlas.

https://www.mhmedical.com

12. Larsen WJ. Human Embryology.

https://www.elsevier.com

13. Avery's Diseases of the Newborn. Elsevier.

https://www.elsevier.com

14. American Academy of Pediatrics (AAP). Neonatal guidelines. https://www.aap.org

15. Sadler TW. Embryology principles and developmental biology. https://shop.lww.com