

RISK FACTORS CAUSED BY CONGENITAL DISORDERS IN CHILDREN

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ABSTRACT

Although some birth defects can be controlled and treated, approximately 3,2 million of these children are lifelong disabled. In addition, birth defects are a leading cause of infant mortality. But where do these shortcomings come from? While some congenital defects are inherited, others are the result of harmful environmental factors known as dermatogens, while others are the result of complex interactions of genetic and environmental influences. But in about half of cases of birth defects, the causes are unknown. Despite the progress made in further deepening the reform of the health care system in our country, strengthening and protecting the health of children, congenital anomalies (developmental defects) play a leading role in the structure of their morbidity, disability and mortality. Among them, 7 500 are said to be infants under one year of age. 60,5% of infant deaths were due to perinatal cases, 16,8% to respiratory defects, 11,7% to congenital anomalies and 11% to other diseases.

KEYWORDS: *Congenital Anomalies, Congenital Malformation, Congenital Heart Defects, Perinatal Causes*

INTRODUCTION

Globally, congenital anomalies prevalent among the population are an important medical and social problem. They have doubled in the last decade of the twentieth century and have played a significant role in the pathology and mortality of infancy, perinatal and childhood. Today, 1,800 out of 10,000 children worldwide are born with congenital anomalies. [1,2]

The emergence of developmental anomalies has been of interest to scientists for several years. However, the etiology and pathogenesis of this complex problem are still insufficiently studied. With this in mind, the appearance of defects in the fetus can be either due to a mutation, or due to the teratogenic effect of some factor, or a combination of the two. [3,4]

The causes of birth defects in humans can be thought of as follows: endogenous causes:

1. Hereditary changes
2. endocrine diseases
3. "Maturation" of germ cells
4. The age of the parents

Exogenous causes:

1. Physical factors:
 - a) radiation
 - b) mechanical effects
2. Chemical factors:
 - a) drugs
 - b) chemicals used in industry and household
 - c) hypoxia
 - d) poor quality food
3. Biological factors:
 - a) viruses
 - b) mycoplasmas
 - c) protozoal infections

In addition, it is necessary to recognize some pathological conditions in the mother's body, the negative impact on fetal development. Among these reasons is Saveleva G. According to M et al, the most common are satellite insufficiency. [5,6]

One of the main factors of perinatal pathology in Uzbekistan is the presence of anemia in pregnant women.

Endogen causes: The most important of these factors, and the worst in terms of consequences, is a mutation in heredity. According to some scientists, almost all developmental defects in humans are the result of mutations. Mutations can occur at three levels: gene, chromosome, and genome.

The importance of the fact that most hereditary-related birth defects are caused by mutations in genes can be clearly seen in pregnancies that end in abortion. In humans, mutations occur as a result of simple physiological processes (natural mutagenesis) or as a result of additional effects on hereditary structures (physical, chemical, biological) (induced mutagenesis). The most dangerous of the physical mutagens now is ionizing radiation. From a large number of chemical mutagens to insecticides, fungicides and herbicides widely used in agriculture, important in clinical teratology, in industry of which formaldehyde, acrolein, epoxides, benzene, arsenic, aromatic hydrocarbons used in the food industry, and 26 others can be shown, these substances can have mutagenic effects from the germ cell to the developing fetus. [7,8]

Numerous observations show that the chromosomes of somatic cells are damaged by viruses of diseases such as epidemic hepatitis, influenza, measles, chickenpox, typhoid. Another endogenous factor is hormonal changes and metabolic defects that occur in pregnant women, which often result in involuntary abortion of the fetus. Sometimes, the morphological and functional differentiation of fetal organs is disrupted, and such babies die within a very short time after birth. It has been proven for endocrine diseases such as diabetes, endemic cretinism, and pathological conditions such as phenylketonuria, galactosemia, and histidinemia. Among them, the most important in the clinic are fetal injuries of women with insulin-dependent diabetes mellitus and phenylketonuria. Examples include diabetic embryopathy and diabetic phenopathies. Babies born with phenopathy are more prone to overweight. It is observed in infants' chest, increased subcutaneous fat layer, fatty dystrophy of the liver, hyperplasia of the pancreatic islets, decreased glycogen accumulation in the myocardial muscles, liver, microangiopathies. [9,10]

In diabetic embryopathies, defects occur in the musculoskeletal, cardiovascular, and central nervous systems. The mechanism of occurrence of these defects is associated with hypoglycemia, hypoxia, vascular disorders, and finally changes in fat and amino acid metabolism. [11,12]

The health of the new generation is related to the health of mothers. The reproductive function of an organism is governed by general biological laws. Periods of development, puberty, and extinction are, of course, reflected in the new generation, in other words, it has been proven many times that parents who have not yet reached the age of puberty are beginning to experience various defects in the generations. Children born to young mothers are more likely to have defects of the limbs and respiratory system, while children born to mothers over the age of 35 are more likely to have defects, especially central nervous system disorders. As parents age (especially mothers), the number of children born with chromosomal disorders increases. The increase in birth defects in children of older parents is due to some factors. [13,14]

Exogen factors: Opinions about the teratogenic effects of X-rays and the effects of other ionizing rays on fetal development vary. It is difficult to make a comprehensive analysis of this issue, of course. It should be noted that the teratogenic properties of ionizing radiation depend on the stages of fetal development, its dose, the intensity of the type of light, as well as the individual sensitivity of the organism. It is known that the nervous system is most sensitive to the

damaging effects of rays. Direct exposure of the fetus to ionizing light can lead to defects in the fetus as a result of metabolic disorders in the pregnant woman, disruption of enzymatic processes, changes in the permeability of cell membranes. Therefore, as mentioned above, the central nervous system and other related (microcephaly, hydrocephalus) defects occur. When it comes to the effects of chemical factors, all scientists agree that any new chemical, even medication, should not be administered as much as possible during the first trimester of pregnancy (until a clinical trial). When evaluating the teratogenicity of chemical factors, of course, it is necessary to take into account the developmental stage of the embryo, genetically sealed susceptibility, the state of the mother organism, the chemical structure of the substance, its ability to pass through the placenta, metabolism. [15,16]

The nature of the transit of chemicals depends on the mass of their molecules, the solubility in lipids, the properties of their binding to serum proteins. It turns out that chemical compounds with a molecular mass greater than 1,000 cannot pass through an undamaged satellite. Thus, these substances do not have teratogenic properties under normal conditions because the chemical acts directly on the embryonic cells. As for the lack of satellite permeability, it is relative, and as a result of an increase in the concentration of a substance or drug in the mother's blood, the substance can pass from this natural barrier to the fetus. It is important how the substance or drug is delivered to the body, in other words, whether in small doses, or repeatedly, or in large quantities over a short period of time. Chronic consumption of some drugs stimulates the formation of enzymes involved in their metabolism, as a result of which these drugs are eliminated from the body and have no teratogenic effect. It is also important how substances enter the body. For example, oral administration of drugs results in their rapid breakdown and inactivation. Drugs of the tranquilizer group that have teratogenic effects for human embryos include thalidomide and diazepam. [17,19]

Babies born to pregnant women who have taken thalidomide develop a variety of defects in the limbs, gastrointestinal and nervous systems. Drugs that prevent seizures also have teratogenic properties, leading to folic acid deficiency in the fetus. [19,20]

Typical symptoms for embryopathies associated with taking anticonvulsants include microcephaly, palate, nose, ears, upper lip, and other defects. Later, psychomotor developmental delay and mental retardation are observed in such infants. [21,22]

As a result of taking anticoagulant drugs in the early stages of pregnancy, hypoplasia of the nose, damage to the optic nerve and developmental delay are observed in the developing fetus. [23,24]

According to some data, treatment of pregnant women with penicillin can lead to the birth of a child with elastosis (skin damage). Thus, some drugs can have both teratogenic and mutagenic effects. [25,26]

Among the substances that adversely affect fetal development, alcohol is of practical importance. There is a lot of information in the medical literature about alcoholic embryopathies. These congenital defects are characterized by hypoplasia, slowing of physical and mental development after birth, and various other defects. The most serious changes are mainly manifested in the development of the brain. [27,28]

L.O. Badalyan and E.M. Mastjukova accurately described the features of the development of fetal alcohol syndrome. Speech development of such newborns slows down, there is a lack of

thinking. In addition, various psychosomatic disorders can occur. The development of this syndrome is associated with a decrease in folic acid in the embryonic and fetal tissues. This is due to the long-term circulation of ethanol in the blood of partially decomposed products, in particular, acetyldehyde. In addition, it should be noted that the activity of fetal alcohol-degrading alcohol dehydrogenase is 10%. [29,30]

Gasoline, benzene, formaldehyde, lead, mercury particles, and many other chemicals that are widely used in industry and agriculture can have embryotoxic effects or cause birth defects.

CONCLUSION

Improving the efficiency, quality and popularity of health care, as well as the formation of a system of medical standardization, the introduction of high-tech methods of diagnosis and treatment, the promotion of healthy lifestyles and disease prevention through the creation of effective models of patronage and dispensary case, approached with special responsibility.

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