

PREVENTIVE MEASURES FOR HEREDITARY DISEASES

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Abstract. Every individual wishes to leave behind healthy offspring. The deterioration of the ecological environment, both in Uzbekistan and worldwide, negatively affects the birth of healthy children. The severe course of hereditary diseases, their frequent fatal outcomes, the absence of effective treatment methods for many of them, and their transmission from generation to generation make the development of preventive measures an issue of great importance.

If young people seek medical genetic counseling before or after marriage or during pregnancy, they significantly increase their chances of having healthy children. The primary goal of medical genetic counseling centers is to prevent the birth of children with hereditary diseases.

This article examines the types of prevention of hereditary diseases, medical genetic counseling, prenatal diagnosis, the consequences of consanguineous marriages, as well as prospects in the treatment and prevention of hereditary diseases. It highlights current achievements in medicine and ongoing challenges in this field.

Keywords: segregation, prevention, prospective counseling, retrospective counseling, non-invasive, invasive, amniocentesis, chorionic biopsy, cordocentesis, placentocentesis, fetoscopy, replacement therapy, vitamin therapy, diet therapy, surgical treatment.

Prevention of Hereditary Diseases

Types of Prevention of Hereditary Diseases

The great Russian writer Leo Tolstoy began his novel Anna Karenina with the famous words: "All happy families resemble one another; each unhappy family is unhappy in its own way." This idea can also be applied to families with healthy or sick children, since each family affected by hereditary disease experiences its own unique form of misfortune.

All hereditary pathologies are associated with a genetic "burden" transmitted from generation to generation. This burden has two main components:



1. **Segregation**, that is, the transmission of a pathological gene from parents to children;
2. **Newly formed mutations**, resulting from changes in the hereditary material of parental germ cells, leading to the birth of affected children from healthy parents.

The severe clinical course of hereditary diseases, their frequent fatal outcomes, the lack of effective treatment methods for many of them, and their inheritance patterns make the development of preventive strategies extremely important.

Prevention is a set of measures aimed at preventing the occurrence and progression of hereditary diseases. Currently, four main types of prevention are widely used.

Primary Prevention

Primary prevention is based on family planning and includes three main directions:

- a) Optimal reproductive age is considered to be between 21 and 35 years. Childbirth before or after this age increases the risk of hereditary pathologies;
- b) If a family has a hereditary disease with a high genetic risk and effective prenatal diagnostic methods are unavailable, it is recommended to refrain from having children;
- c) In cases where both partners are heterozygous carriers of the same hereditary disease and the genetic risk is high, reproduction is discouraged. Reducing consanguineous marriages is one of the most important measures in preventing hereditary diseases.

Secondary Prevention

Secondary prevention focuses on environmental protection through the development and enforcement of sanitary and hygienic standards aimed at reducing mutagenic factors in the human environment. These include mutagenic drugs, X-rays, radioactive and magnetic radiation, chemical food additives, nitrites, and other harmful substances.

Tertiary Prevention

Tertiary prevention is based on prenatal diagnosis. The use of prenatal diagnostic methods has significantly reduced the birth rate of children with hereditary diseases worldwide over the past decade. Determining fetal sex in utero plays a crucial role in preventing sex-linked hereditary disorders. Amniotic fluid analysis for sex chromatin (performed at 18–20 weeks of gestation), maternal blood lymphocyte testing, and hormonal analysis (testosterone levels) are commonly used. If severe hereditary pathology is detected, termination of pregnancy may be considered.

Quaternary Prevention

Quaternary prevention involves regulating gene expression through phenotypic correction of pathological gene effects. Intervention can occur at different stages of ontogenesis. For example, dietary and pharmacological treatment of pregnant women may correct certain fetal abnormalities. Early vitamin supplementation during pregnancy has been shown to reduce the risk of cleft lip and palate.

Postnatal correction of metabolic disorders such as galactosemia and phenylketonuria through dietary therapy is widely practiced. Limiting exposure to triggering factors (e.g., avoiding



sulfonamides or smoking cessation) can prevent complications such as early-onset pulmonary emphysema.

Medical Genetic Counseling

Medical genetic counseling plays a crucial role in identifying, preventing, and managing hereditary diseases. It aims to inform families about disease risks, inheritance patterns, and preventive options. Counseling may be **prospective**, conducted before the birth of a child at risk, or **retrospective**, carried out after the birth of an affected child.

Genetic risk assessment is based on theoretical calculations, empirical data, or a combination of both. Risk levels are classified as low (up to 5%), moderate (6–10%), intermediate (11–20%), or high (above 21%). Counseling decisions consider not only risk levels but also disease severity, treatability, and quality of life.

Prenatal Diagnosis

Prenatal diagnosis involves examining the fetus before birth to detect hereditary abnormalities. Non-invasive methods include ultrasound, biochemical, and molecular genetic tests. Invasive methods include amniocentesis, chorionic villus biopsy, cordocentesis, placentocentesis, and fetoscopy.

Amniocentesis is the most commonly used method, allowing cytogenetic, biochemical, and molecular analysis of fetal cells. Alpha-fetoprotein levels in amniotic fluid and maternal blood help detect neural tube defects.

Consequences of Consanguineous Marriages

Consanguineous marriages significantly increase the risk of hereditary diseases due to the higher likelihood of recessive gene expression. Such unions are common in certain regions, including parts of Central Asia. Studies show higher rates of stillbirths, congenital anomalies, and intellectual disabilities among children born from consanguineous marriages.

Preventive measures include mandatory premarital genetic counseling and public awareness programs.

Treatment of Hereditary Diseases

Although hereditary diseases are often considered incurable, many can be managed effectively. Treatment methods include:

1. Replacement therapy;
2. Vitamin therapy;
3. Diet therapy;
4. Surgical treatment.

Early diagnosis significantly improves outcomes by preventing irreversible organ damage.



Conclusion

In conclusion, preventive measures for hereditary diseases are of great importance for the future of humanity. Research and practical efforts in this field contribute significantly to disease prevention, treatment, and the reduction of genetic disorders in future generations. Continued development of medical genetics will help ensure healthier families and societies.

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