Prevention of hereditary diseases

The nation's gene pool is its strategic resource.

The gene pool primarily determines the physical and mental health of people, their performance, resistance to stress, intellectual and cultural potential.

The task of preserving the gene pool is recognized as a necessary condition for the prosperity of the nation.

The gene pool is the result of the interaction of the genome with the environment.

It is not so much genes that are important as signs (hair dryers).

The pathological gene may not manifest in the phenotype if there are no suitable conditions for this.

The action of genes can be controlled!

The total social effect with a total life expectancy of 70 years is presented:
Uncomplicated life;
Complicated life;
A shortened life.

For patients with hereditary pathology:
Uncomplicated life - 20 years;
Complicated life - 31 years;
Shortening of life - for 19 years.

Social adaptation in individuals with hereditary diseases, their intellectual and physical development are sharply reduced.

Almost 3/4 of children have a low level of ability to learn in school or to work.

The structure of disability in a variety of hereditary diseases(%)

Categories invalidism	AD	AR	X-linced	All
Soft	39,4	12,6	14,3	25,8
Big	59,9	73,2	78,3	67,2
Incompatible with life	0,7	14,2	7,1	7,0

The purpose of prevention of hereditary human pathology is to prevent the emergence of new mutations and the transmission of existing mutational disorders to the next generations.

Mutational load-a set of mutational changes that occur in the genetic material of a person (individuals of one generation) throughout life under the influence of mutagens habitat.

Segregation of the cargo – is genetic defects that are inherited by people of one generation from the representatives of the other (previous) generation.

Prevention of genetic cargo

Type 1 prevention

prevention of conception of a sick child: medical and genetic counselingteratology counselingpericonceptional prevention treatment of chronic pathology of women Definition of Rh-belonging in spouses

Regulation of optimal reproductive age (for women 21-35 years old)

Type 2 prevention

prevention of birth of children with hereditary pathology:termination of pregnancy in case of prenatal diagnosis of hereditary pathology.Medical abortion or preterm birth can be carried out only with the consent of the woman and within a strict time limit.

Type 3 prevention

tactics of management of newborns with vnz:it is aimed at ensuring the non-manifestation (minimization) of the pathological phenotype, as well as social adaptation of the child

There are two organizational approaches to the prevention of hereditary pathology:

- family (carried out through medical and genetic counseling)
- populational

Population-based prevention

special state screening programs for any hereditary pathology or heterozygous carrier of mutant genes-sanitary and hygienic measures to identify harmful production factors and adverse environmental influences

Ways of prevention and their implementation:

1. Fortification of women before conception and in the first months of pregnancy reduces the frequency of HRV, diet therapy for gene diseases, correction of environmental factors in multifactorial pathology (MFP).
2. Elimination of embryos and fetuses with hereditary pathology-artificial termination of pregnancy. This path is created by evolution in the form of spontaneous abortions.
3. Planning of childbearing in medical and genetic counseling. Extremes must be avoided.
4. Protection of the human environment in all its aspects.

Organizational forms of prevention of hereditary diseases:

- ✓ diagnosis of heterozygous carrier;
- ✓ prenatal diagnosis;
- ✓ medical and genetic counseling;
- ✓ prophylactic medical examination;
- ✓ test of mutagenicity;
- ✓ hygienic regulation;
- ✓ promotion of medical and genetic knowledge.

Prevention of hereditary pathology

Principles:

- ✓ Medical and genetic counseling for families with sick children or relatives suffering from congenital or hereditary diseases.
- ✓ Conducting mass screening programs to prevent the birth of a child with congenital pathology or detection of preclinical forms of the disease in newborns.
- ✓ Prenatal diagnosis using modern research methods to identify developmental abnormalities, congenital or hereditary diseases in the fetus.
- ✓ Clinical examination of families in medical and genetic counseling with genetic disorders, follow-up study of the proband and his family.
- ✓ Reducing the impact of mutagenic environmental factors.

Prevention of hereditary pathology
Levels:

- ✓ pregametic,
- ✓ prezygotic,
- ✓ perinatal,
- ✓ postnatal.

Pregametic level

It is based on the improvement of the population of reproductive age, especially women:

- sanitation of foci of infection, fortification of the body,
- observance of a healthy lifestyle, exclusion of exposure to mutagenic factors during life,
- the usage precautions of the spouses prior to conception,
- exclusion of teratogenic environmental factors, exacerbation of chronic diseases or infection of the body.

Prenatal level

Methods of prenatal diagnosis aimed at preventing the birth of a child with hereditary or congenital pathology.

Postnatal level

Preventive screening in newborns for early preclinical detection of monogenic forms of hereditary diseases.

Types and terms of diagnostics:

Noninvasive:

- Ultrasound diagnosis-10-12,20-24, 30 weeks.,s
- creening for AFP, HCG - 16-20 weeks.

Invasive:

- transcervical chorionic villus sampling – 9-11 weeks.,
- transamniotic chorion biopsy - 9-11 weeks.,
- transamniotic amniocentesis-after 15 weeks.,
- transamniotic cordocentesis-after 18 weeks.

Invasive prenatal diagnosis

Indications:

- ✓ Age women 35 years and higher.
- ✓ Repeated modified data of pregnant women screening for AFP, HCG.
- ✓ Cases of birth in the family of children with congenital malformations.
- ✓ Passage of x-ray diagnostic procedures by pregnant women in the early stages.
- ✓ The presence in the history of pregnant women indications of infectious viral diseases transferred in the early stages of pregnancy.
- ✓ The application directly before pregnancy or during its development of teratogenic drugs.
- ✓ The presence of hereditary disease in one or both parents.

Invasive prenatal diagnosis

Indications:

- ✓ The presence of balanced rearrangements in one or both parents.
- ✓ Establishment of ultrasound markers of chromosomal pathology of the fetus.
- ✓ Contact of a pregnant woman with harmful industrial and adverse environmental factors.
- ✓ Karyotyping of the fetus at the termination of this abnormal pregnancy (to establish the prognosis for the next pregnancy).

Medical and genetic counseling

this is a type of specialized medical care, as a result of which patients and their relatives with the risk of hereditary disease receive information about the consequences of this disease, the probability of its development and inheritance, as well as ways to prevent it.

It appears in medical-genetic consultations.

Medical and genetic counseling

Tasks:

- ✓ Determination of health prognosis for future offspring in families where there was or is a patient with hereditary pathology.
- ✓ Explaining to parents in an accessible form the meaning of genetic risk and assisting in making the right decision.
- ✓ Help doctors in the diagnosis of hereditary disease, if special genetic methods are required.
- ✓ Promotion of medical and genetic knowledge among health workers and the population.

To detect hereditary pathology it is necessary:

- ✓ Data should be collected clinical and genealogical method, to perform the necessary examination and differential diagnosis.
- ✓ Pay attention to recurrent, chronic, long-term untreatable diseases, especially in children.
- ✓ Identify specific symptoms or syndromes.

To detect hereditary pathology it is necessary:

Pay attention tothe involvement of many systems and organs (presistemnomu lesions, the presence of congenital defects and micro anomalies of development of dysplasia).

To find out the congenital nature of the pathology.

Dysplasia development, impaired development of tissues, organs or systems of the body without disruption of their functions (in contrast to malformations).

The presence of five or more developmental dysplasias in a person allows the doctor to suspect hereditary pathology or hidden congenital malformations.

Empirical risk

Craniocerebral hernia:
2-5% after the 1st affected,
10% after the 2 nd affected,
20% after the 3rd affected.
Hypertrophic pyloric stenosis:
if the proband is male – 6% for sons,
and 2% for daughters,
if the proband is female – 20% for sons,
7% for daughters.
Schizophrenia if ill:
one of the parents – 10%,
both parents are 40%,
one of the proband sibs (sporadic case) is up to 20%.

Indications in obstetrics and gynecology:

- ✓ Primary infertility of the spouses, if we exclude gynecologic pathology women and abnormal spermatogramma men.
- ✓ Primary amenorrhea with underdevelopment of secondary sexual organs.
- ✓ Repeated spontaneous abortions, stillbirths after exclusion of gynecological pathology.
- ✓ Birth of a child with developmental disabilities.
- ✓ The presence of any hereditary disease in a pregnant woman or her husband.
- ✓ Incompatibility of RH and ABO spouses if there is a history of RH conflict or ABO conflict in previous pregnancies.
- ✓ Work of a pregnant woman on harmful production.
- ✓ Large doses of drugs that were given to spouses before conception or in the first months of pregnancy.
- ✓ Mental retardation-isolated and combined with pathology of other organs and systems.
- ✓ Other disorders of mental status, such as disinhibition or autism, behavior defects, dyslexia, alalia, decreased intelligence, memory.

Indications in Pediatrics:

- ✓ Chronic lung diseases-recurrent bronchitis, lung abscesses.
- ✓ Violation of physical development of children-growth retardation, deformation of bones, trunk and limbs, excessive deposition of fat, stiffness or looseness of joints, decreased vision, blindness, hearing loss and deafness, impaired sense of smell.
- ✓ Seizures, muscle Hypo-and hypertension, gait and coordination disorders, skin rash, Hypo - and hyperpigmentation, photosensitivity, jaundice.
- ✓ Intolerance to certain foods and drugs, digestive disorders: frequent vomiting, diarrhea, loss of appetite, fatty stools, hepatomegaly, splenomegaly, gingivitis.
- ✓ Chronic diseases of unknown origin, poorly treatable.
- ✓ Intolerance to certain types of food and drugs.
- ✓ Family forms of allergoses with varying degrees of manifestation-bronchial asthma, vasomotor rhinitis, eczema, urticaria, etc.
- ✓ Kidney stone disease in children, unusual color and smell of urine.
- ✓ Hemolytic anemia.

Indications in the clinic of internal diseases:

- ✓ Gastric ulcer and 12 duodenal ulcer.
- ✓ Hypertension and atherosclerosis.
- ✓ Blood disease / hemolytic anemia, hemophilia, Werlhof disease, etc.
- ✓ Metabolic disorders and endocrine diseases (diabetes, toxic goiter, obesity, etc.)
- ✓ Family nephropathy.
- ✓ Defeat of the musculoskeletal system (dwarfism, Marfan syndrome, pseudoachondroplasia, etc.)

Periconception prevention

- A set of measures potentially capable of providing optimal conditions for the maturation of the egg, its subsequent development, implantation, and as a result – the development of the fetus.
- Preconceptional prevention is carried out in relation to congenital malformations and other multifactorial conditions, i.e., not determined by mendeliruyuschim inheritance. Prevention is carried out after a medical genetic study of the family, during which the geneticist determines the nature of the inheritance of the disease, the repeated genetic risk, the possible effectiveness of prevention.

Periconceptional prevention is possible only through planned pregnancy:

- for 3-4 months. before possible conception, future parents undergo a detailed medical examination;
- after that, the couple receive preventive treatment for 2-3 months;
- before the planned conception.

Within 2-3 months. pregnant woman receives certain drugs

Complex periconceptional prevention includes:

1. Treatment of chronic foci of infections (if any) in expectant parents.
2. Treatment of chronic somatic diseases.
3. Evaluation of the semen.
4. Regulation of the sexual mode, planning to become pregnant.
5. A diet enriched with vitamins and trace elements, including folic acid (it is believed that it reduces the risk of having a child with CNS defects).

Indications for periconceptional prophylaxis (Hollingsworth):

- ✓ genetic risk of multifactorial malformations in the family;
- ✓ repeated spontaneous abortions and the birth of dead fetuses;
- ✓ birth of children with prenatal hypotrophy and preterm birth history;
- ✓ diabetes mellitus, other endocrine and metabolic diseases in the mother;

Indications for periconceptional prophylaxis (Hollingsworth):

- ✓ chronic somatic diseases in one or both parents;
- ✓ occupational hazards in one of the spouses;
- ✓ eating disorders;
- ✓ long-term use of drugs;
- ✓ diseases caused by TORCH-infections.

TORCH complex – WOH, 1971)

Group of 4 viral and bacterial infections :

To - toxoplasmosis,

R - rubella,

C – cytomegalovirus infection,

H – herpes.

Chlamydia, gonococcal infection, trichomoniasis, genital Mycoplasma, syphilis and HIV do not belong to TORCH infections.

The program of screening and management of pregnant women when they are detected is different from the screening and management of pregnant women with TORCH infections.

Toxoplasmosis

About 10 -12 % of women are infected with toxoplasmosis during pregnancy.
The risk of infection to the fetus is 30-40 % .
If the infection occurred in the I, II trimesters of pregnancy, the risk of spontaneous abortion and premature birth reaches 10-15 %.
With congenital toxoplasmosis up to 15-25%, severe fetopathies are noted: microcephaly, hydrocephalus, chorioretinitis, hepatosplenomegaly, thrombocytopenia.

If the infection occurred in the third trimester of pregnancy, the risk of congenital toxoplasmosis is up to 90 %, but the disease is asymptomatic in most cases.

Rubella

When infected with rubella virus before 16 weeks of pregnancy often occur:

- Intrauterine fetal death
- Macro and microcephaly
- Gregg's triad:

eye damage + deafness + cardiovascular defects

When infected after 16 weeks of pregnancy, the risk of congenital abnormalities is significantly reduced.

Cytomegalovirus infection

Approximately 1% to 4 % of pregnant women are infected with cytomegalovirus during pregnancy, one third of them have intrauterine infection of the fetus.

Infection before 12 weeks leads to miscarriages and congenital deformities :

- Hydro or anencephaly,
- Cerebral palsyLesions of the organs of hearing and vision

Infection after 12 weeks manifests itself as a generalized form of cytomegaly-hepatitis, pneumonia and retinitis.

Herpes simplex type 1/2

Primary infection with herpes simplex virus can lead to:

- Violation of the course of pregnancy-polyhydramnios, termination of pregnancy, miscarriage
- Pathology of the fetus intrauterine infection, spontaneous abortion, stillbirths
- Pathology of the newborn-congenital herpes, prematurity, low birth weight.

With a primary episode of herpes, the risk of transmission to the fetus is up to 50%, with recurrent infection or asymptomatic 0-4%

How to check pregnant women for TORCH infection

Testing of pregnant women for TORCH infections is carried out by detecting antibodies to infections in the blood serum by enzyme immunoassay (ELISA).Detection of "early" class M antibodies and "late" class g antibodies is important.

It is advisable to carry out testing for STICKING infection before the planned pregnancy.

Stages of periconception prevention

Timing of conception (late summer-early autumn).

Examination of spouses before pregnancy (to identify various infectious, endocrine and other diseases).

Taking multivitamin preparations for 2-3 months before conception(folic acid to 0.4-1 mg per day, ascorbic acid, L-tocopherol, b vitamins).

The diet of the spouses is enriched with foods containing folic acid: greens, tomatoes, legumes, liver.

Taking a woman multivitamins to 10-12 weeks of pregnancy.

Monitoring of pregnancy (includes methods of prenatal diagnosis, allowing to detect the disease in the fetus).