

BASES OF MENTAL RETARDATION

ETIOLOGY & PATHOGENESIS OF MENTAL RETARDATION. PART I



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ETIOLOGY OF MENTAL RETARDATION

- endogenous hereditary factors
 - exogenous factors
- РЕПОЗИТОРИЙ БГМУ



MUTATIONS

- **gene mutations**
(changing the structure of genes)
- **genomic mutations**
(chromosome number change)

GENE MUTATIONS

(changing the internal structure of genes)

- **translocation**
(exchange of fragments between chromosomes)
- **deletions**
(breakage of chromosomes with the loss of their fragments)
- **duplications**
(duplication of a chromosome fragment)
- **inversion**
(180 ° rotation of a chromosome fragment)



GENOMIC MUTATIONS

(chromosome number change)



trisomy

(increase of one chromosomes)



monosomy

(lack of one chromosome)

РЕПОЗИТОРИЙ БГПУ

CAUSES OF CHROMOSOMIC MUTATIONS

➤ **physical factors**

(ionizing radiation, electromagnetic radiation)

➤ **chemical factors**

(insecticides, fungicides, herbicides, formaldehyde)

➤ **biological factors**

(viruses, metabolic disorders, maternal age)

RISK OF BIRTH A CHILD WITH A TRISOMY RELATED TO THE AGE OF HIS MOTHER

- 30-34 years – 1 : 500
- 40-44 years – 1 : 60
- > 45 years – 1 : 20

CLASSIFICATION OF BIRTH DEFECTS

- **primary congenital (genocopy)**
genuine microcephaly, Cornelia de Lange syndrome, Down syndrome, trisomy X
- **congenital secondary (phenocopy)**
after-effect of toxoplasmosis, hemolytic disease, hypoxia
- **postnatal acquired**
after-effect of severe intoxication, neuroinfection, cerebral injury, asphyxia

TYPES OF BIRTH DEFECTS

- **gametopathy**

(defeat of the genital cell before fertilization)

- **blastopathy**

(period of blastogenesis lasts up to 4 weeks)

- **embryopathy**

(embryogenesis period lasts from 4 weeks to 4 months)

- **fetopathy**

(period of fetogenesis lasts from 4 months to birth date)

METABOLIC AND ENDOCRINE MOTHER'S DEFECTS

- **phenylketonuria**
(blood phenylalanine > 30 mg / l)
- **diabetes**
(blood glucose > 6 mmol / l)