CHROMOSOMAL NUMERICAL ABERRATIONS

INSTITUTE OF BIOLOGY AND MEDICAL GENETICS OF THE 1ST FACULTY OF MEDICINE
CHROMOSOMAL ABERRATIONS

► NUMERICAL
  - POLYPLOIDY
    - ANEUPLOIDY
      - MONOSOMY
        - TRISOMY
      - TRIPLOIDY
      - TETRAPLOIDY

► STRUCTURAL
  - MOSAICISM

► MIXOPLOIDY
  - CHIMERISM
LIST OF BASIC SYNDROMES

NUMERIC ANOMALIES OF AUTOSOMES:
- DOWN SYNDROME
  47,XX/Y,+21
- PATAU SYNDROME
  47,XX/Y,+13
- EDWARDS SYNDROME
  47,XX/Y,+18

NUMERIC ANOMALIES OF SEX CHROMOSOMES:
- TURNER SYNDROME
  45,X
- KLINEFELTER SYNDROME
  47,XXY
- XYY SYNDROME
  (SUPERMALE) 47,XYY
- XXX SYNDROME
  (SUPERFEMALE) 47,XXX
DOWN SYNDROME
1 : 600 - 800

- hypotonia in newborns
- upslanting palpebral fissures
- neck webbing
- dysplasia of ears
- flat occiput
- single palmar crease
- epicantic folds (inner canthus)
- congenital heart defects, defects of other organs
DOWN SYNDROME

• mental retardation
• macroglossia
• male hypogenitalism
• denture defects
• short, broad hands, brachydactyly
• immune system defects
• higher risk of tumour diseases (leukemia)
PATAU SYNDROME

1 : 15 000 – 20 000

- severe developmental retardation
- congenital heart defects
- microcephaly
- malformed, low-set ears
- microphthalmia
- polydactyly
- kidney anomalies
- cleft palate, cleft lip
PATAU SYNDROME

polydactyly

cleft palate, cleft lip
cyclopaia
EDWARDS SYNDROME

1 : 5 000 - 10 000

- severe developmental retardation
- heart defects
- malformed, low-set ears
- hypoplastic nails
- digits overlapping
- micrognathia
- prominent occiput
- pedes equinovares (clubfoot)
- microcephaly
EDWARDS SYNDROME

- malformed, low-set ears
- pedes equinovares
- micrognathia
- digits overlapping
TURNER SYNDROME

1 : 2 000 – 2 500

- short stature
- gonadal dysgenesis, primary amenorrhoea
- average intelligence
- short webbed neck (pterygium colli)
- low posterior hairline
- broad/shield chest
- palms and feet edema (newborns)
TURNER SYNDROME

HYGROMA COLLI CYSTICUM
- ABORTED FETUS
KLINFELETER SYNDROME
1 : 500 - 1 000

• tall stature
• average intelligence
• male psychosexual orientation
• hypoplastic testes, cryptorchism
• sterility - azoospermia
• gynaecomastia
XXX SYNDROME (SUPERFEMALE)

- 1:1000, no specific phenotype
- average intelligence
- normal sexual development
- decreased fertility (spontaneous abortions), without risk of chromosomal aberrations in offspring
- no increased occurrence of congenital disorders over to population risk
XYY SYNDROME (SUPERMALE)

- „robust“ growth (proportional), especially height
- average intelligence
- normal sexual development
- normal fertility, without risk of chromosomal aberrations in offspring
- controversy – affected psychosocial development

TWINS – TALLER 47,XYY
Enumerate the Barr bodies in:

a) 45,X  a) 0
b) 48,XXXX  b) 3
c) 47,XXY  c) 1
d) 49,XXXXX  d) 4
e) 47,XYY  e) 0
f) 46,XX  f) 1
g) 47,XX,+21  g) 1
h) 48,XXXXY  h) 2
i) 47,XY,+13  i) 0
NONDISJUNCTION

NORMAL

NONDIS. MI

NONDIS. MII
Fill in the scheme of the nondisjunction in meiosis I in mother leading to the birth of a child with Klinefelter syndrome.
Fill in the scheme of the nondisjunction in meiosis I in mother leading to the birth of a child with Klinefelter syndrome.
Fill in the scheme of the nondisjunction in meiosis II in father leading to the birth of a child with Turner syndrome.
Fill in the scheme of the nondisjunction in meiosis II in father leading to the birth of a child with Turner syndrome.
Fill in the scheme of the nondisjunction in meiosis I in one parent leading to the birth of a child with Down syndrome.
Fill in the scheme of the nondisjunction in meiosis I in one parent leading to the birth of a child with Down syndrome
task 12, p.86: A colour-blind mother and father with normal colour vision have a son with normal vision whose karyotype is 47,XXY. Both parents have normal karyotype. In which parent and at which meiotic division did nondisjunction occur?
Nondisjunction occurred during meiosis I in father.
INCREASING FREQUENCY OF THE CHROMOSOMAL ABERRATIONS WITH THE AGE OF THE MOTHER

AGE LIMIT - CHROMOSOMAL ANALYSIS OF THE FETUS IS RECOMMENDED (CZECH REP.)
Prenatal diagnostics

Chorionic villi sample

Amniocentesis (16.-18. w.)

Cordocentesis

FISH (within 24 hours)

Interphase nucleus

Locus specific probes

Karyotype (cca 14 days)

47,XX,+21
47,XXY
45,X
47, XY, +21
47,XX,+13