CHROMOSOMAL NUMERICAL ABERRATIONS

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

CHROMOSOMAL ABERRATIONS

- **NUMERICAL**
  - ANEUPLOIDY
    - MONOSOMY
    - TRISOMY
  - POLYPLOIDY
    - TRIPLOIDY
    - TETRAPLOIDY
- **STRUCTURAL**

- **MIXOPLOIDY**
  - MOSAICISM
  - 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

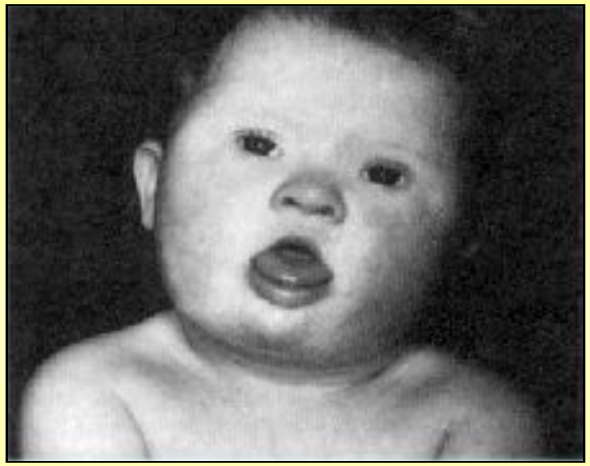
KLINEFELTER SYNDROME
47,XXY

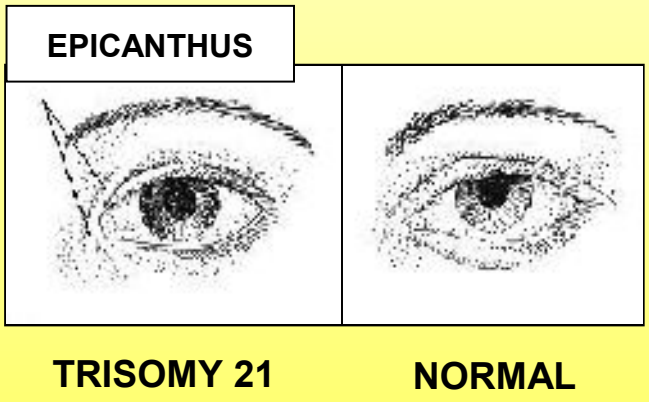
XYY SYNDROME
(SUPERMALE) 47,XYY

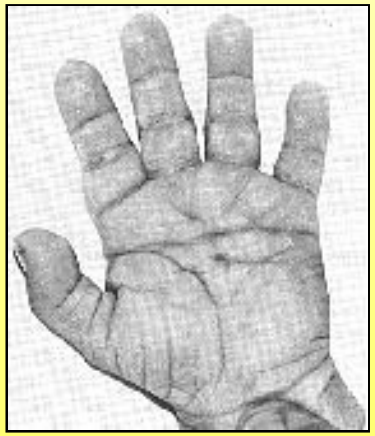
XXX SYNDROME
(SUPERFEMALE) 47,XXX

DOWN SYNDROME

1 : 600 - 800

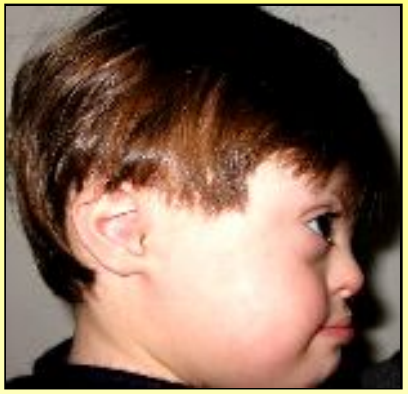


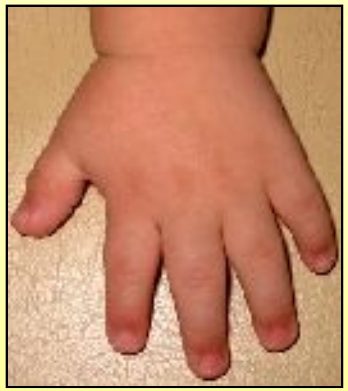


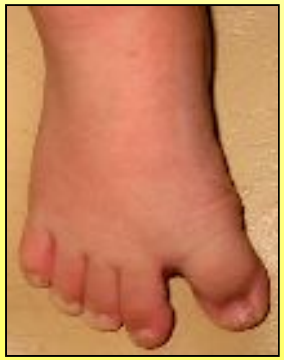


- hypotonia in newborns
- upslanting palpebral fissures
- neck webbing
- dysplasia of ears
- flat occiput
- single palmar crease
- epicantic folds (inner canthus)
- congenital heart defects, defects of other organs

DOWN SYNDROME



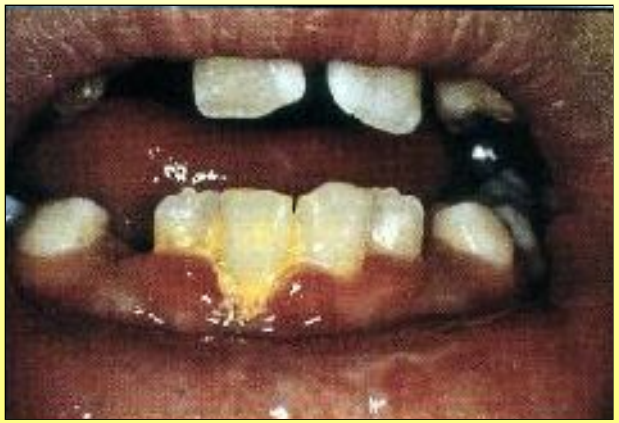




- mental retardation
- macroglossia
- male hypogenitalism
- denture defects
- short, broad hands, brachydactyly
- immune system defects
- higher risk of tumour diseases (leukemia)

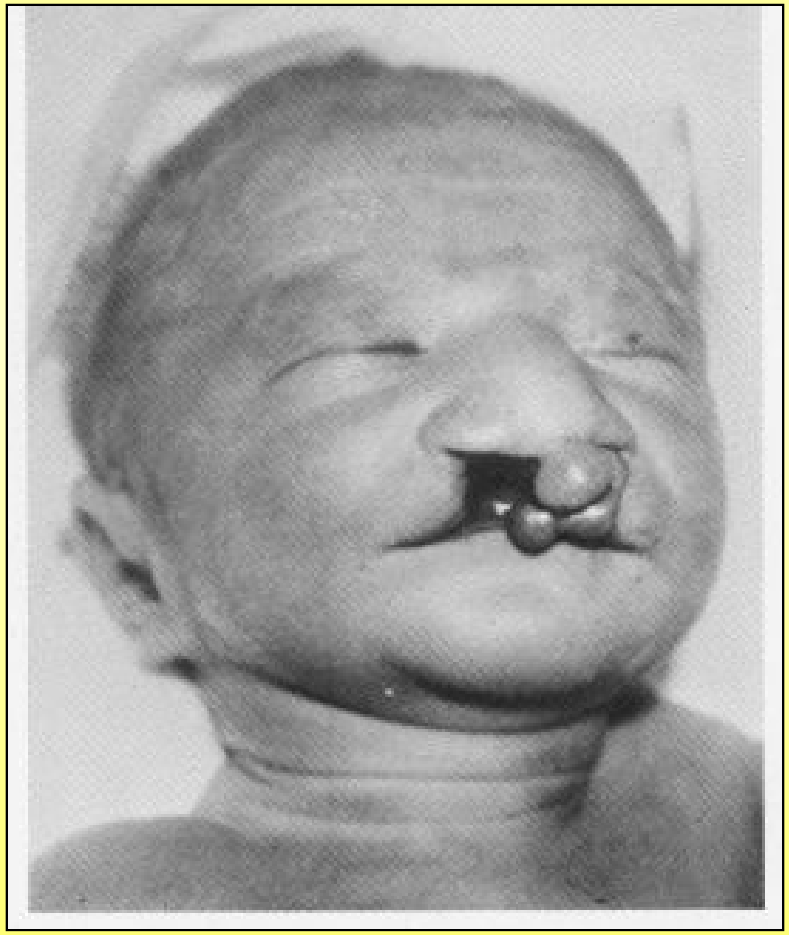
DOWN SYNDROME
DENTAL ABNORMALITIES

- hypodontia – lateral incisors upper and lower, second premolars upper and lower
- changes in tooth size and shape (both permanent and deciduous dentition) – size reduction, shovel-shaped incisors, reduced root lenghts
- increased risk of periodontal disease
- reduced caries prevalence
- underdevelopment of the upper jaw
- delayed eruption of teeth
- third molar agenesis, more often in the maxilla



PATAU SYNDROME

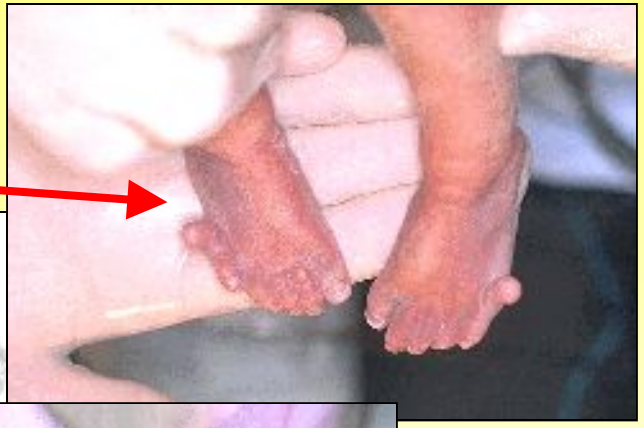
1 : 15 000 – 20 000

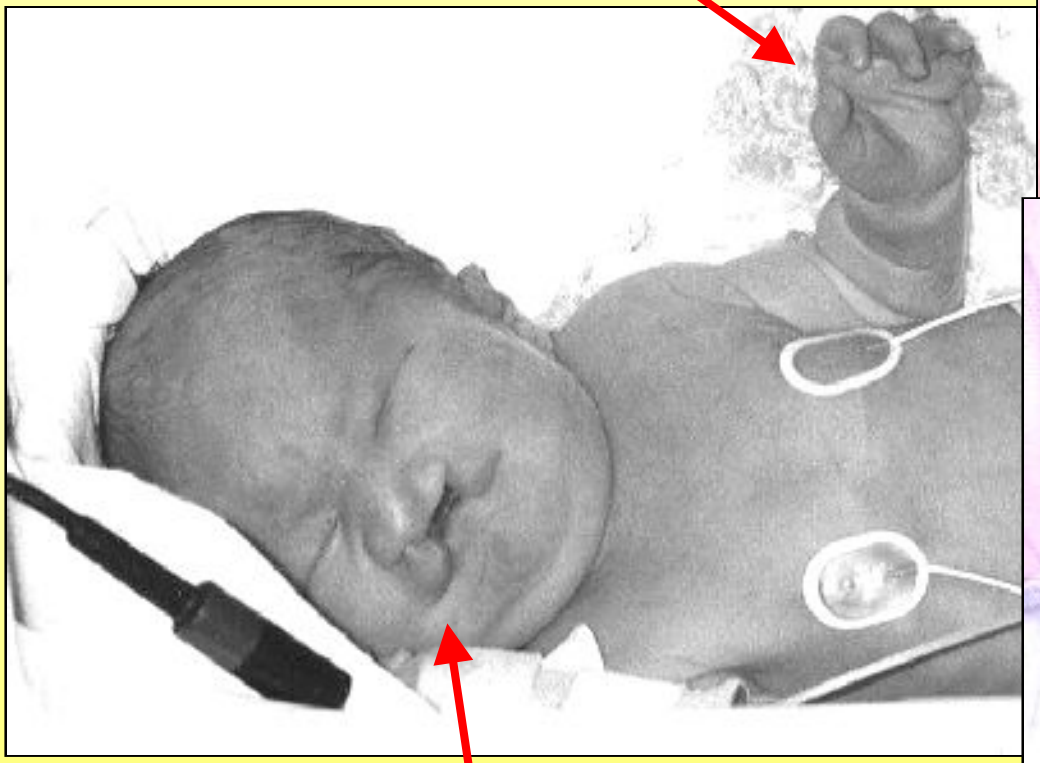


- severe developmental retardation
- congenital heart defects
- microcephaly
- malformed, low-set ears
- microphtalmia
- polydactyly
- kidney anomalies
- cleft palate, cleft lip

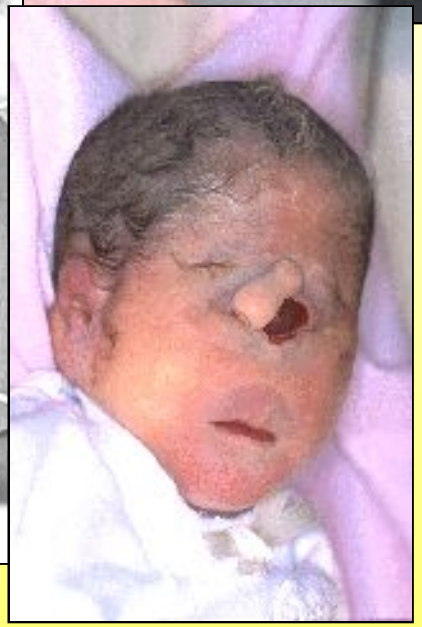
PATAU SYNDROME

polydactyly





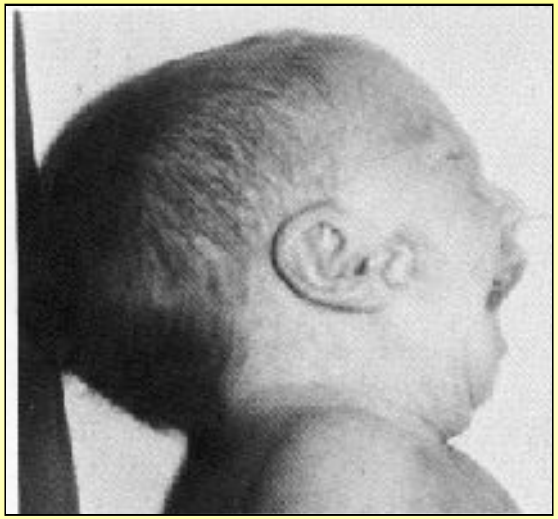
cleft palate, cleft lip



cyclopia

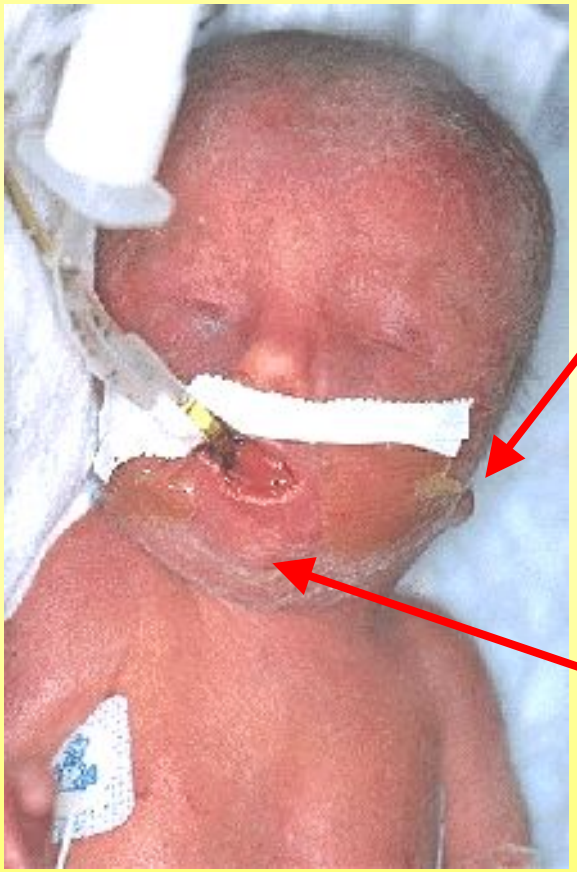
EDWARDS SYNDROME

1 : 5 000 - 10 000



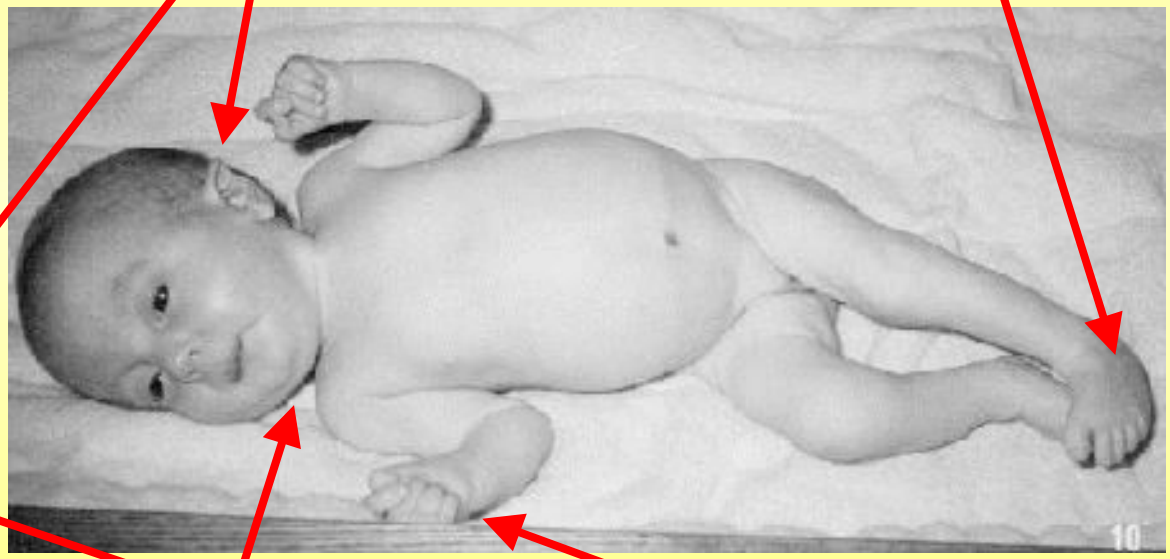
- severe developmental retardation
- heart defects
- malformed, low-set ears
- hypoplastic nails
- digits overlapping
- micrognathia
- prominent occiput
- pedes equinovares (clubfoot)
- microcephaly

EDWARDS SYNDROME



malformed, low-set ears

pedes equinovares

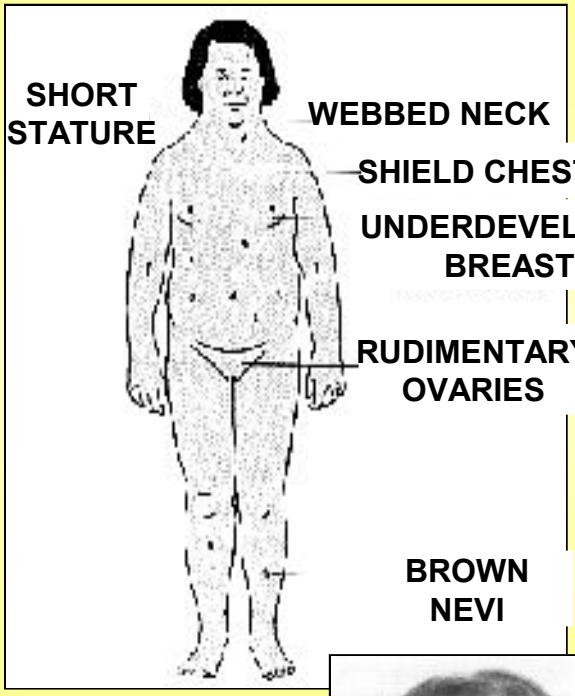


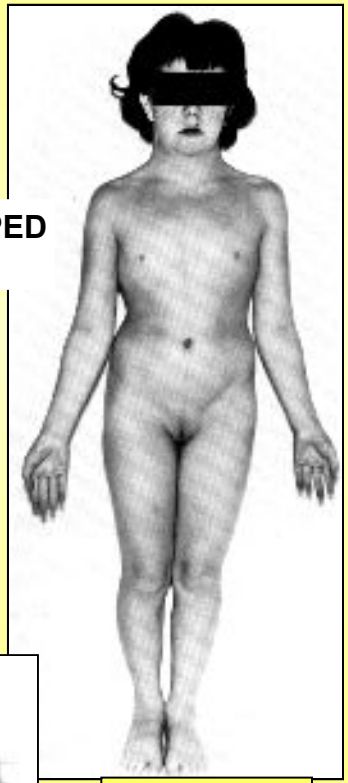
micrognathia

digits overlapping

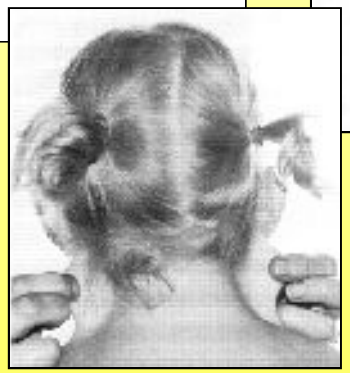
TURNER SYNDROME

1 : 2 000 – 2 500





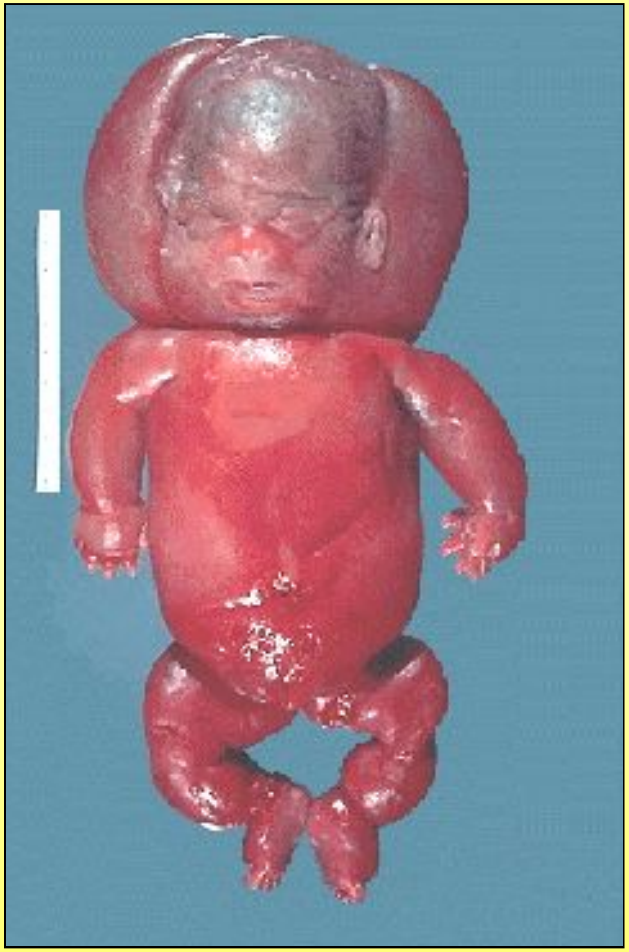
18 YEARS



PTERYGIUM COLLI

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

TURNER SYNDROME



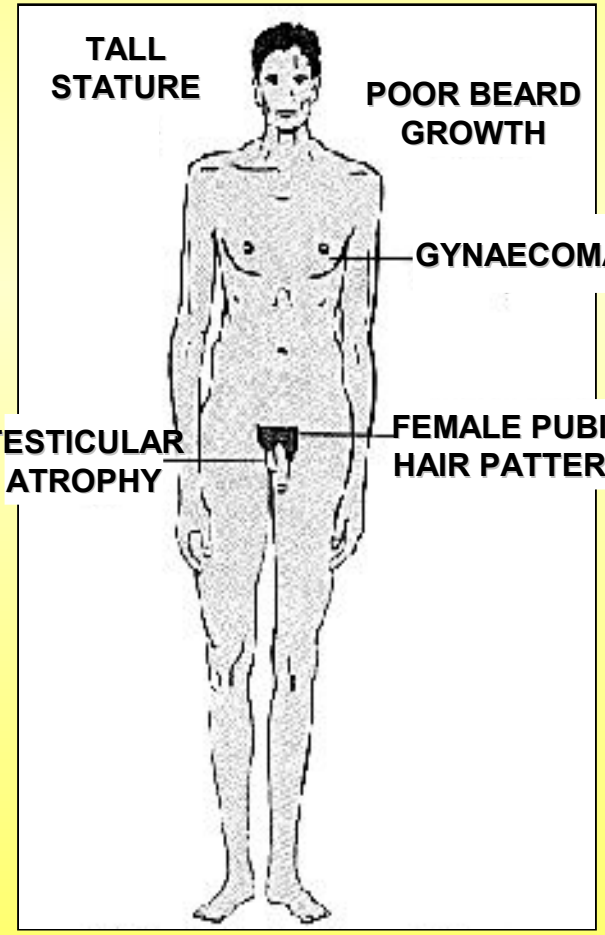
Hygroma colli cysticum (aborted fetus)

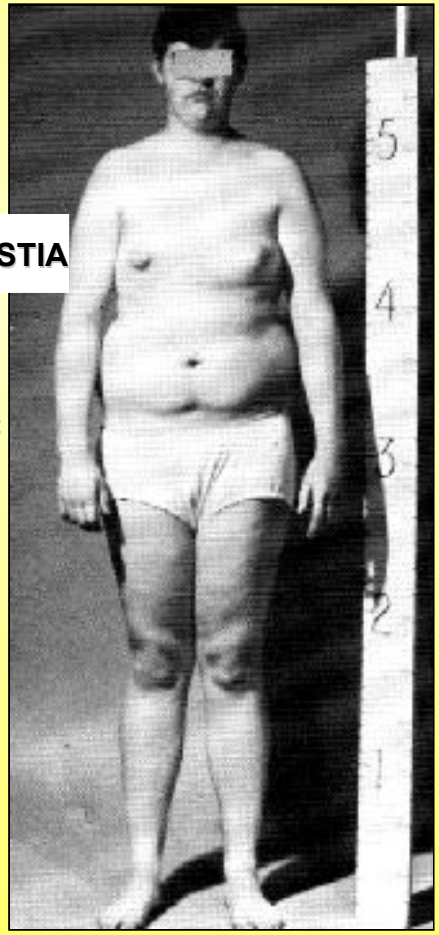
DENTAL ABNORMALITIES

- premature eruption of permanent teeth
- root resorption
- changed palate morphology – high arched palate
- malocclusion

KLINEFELTER SYNDROME

1 : 500 - 1 000



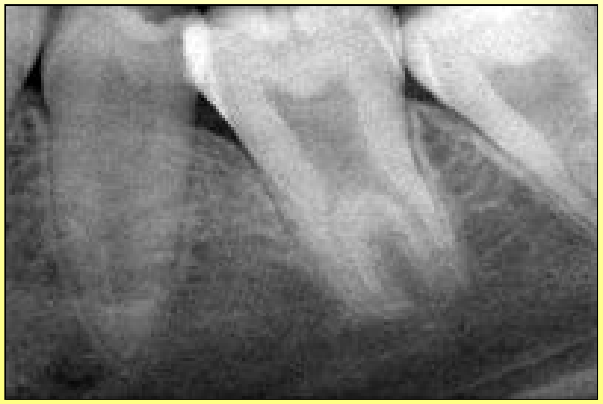


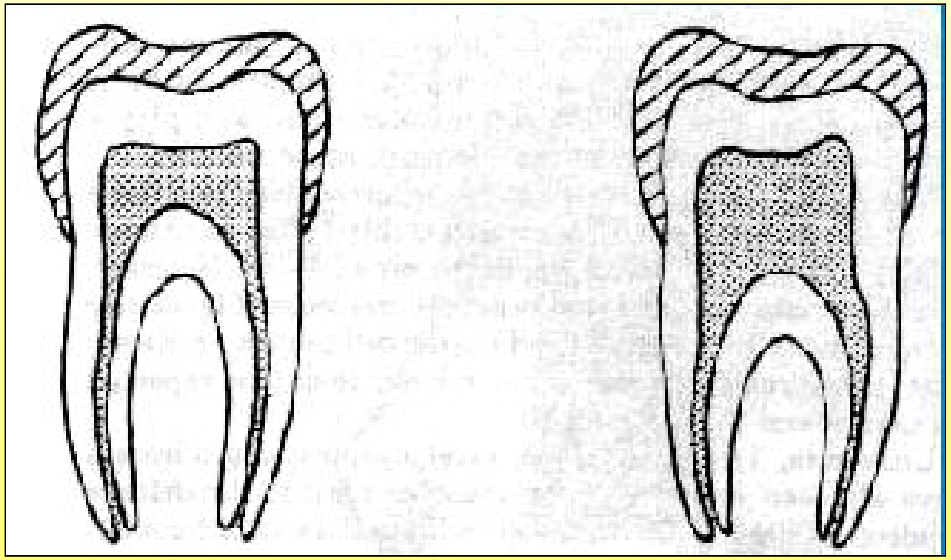
- tall stature
- average intelligence
- male psychosexual orientation
- hypoplastic testes, cryptorchism
- sterility - azoospermia
- gynaecomastia

KLINEFELTER SYNDROME

DENTAL ABNORMALITIES

- taurodontism of molars (enlarged pulp chamber and lengthened crown)
- shovel-shaped incisors





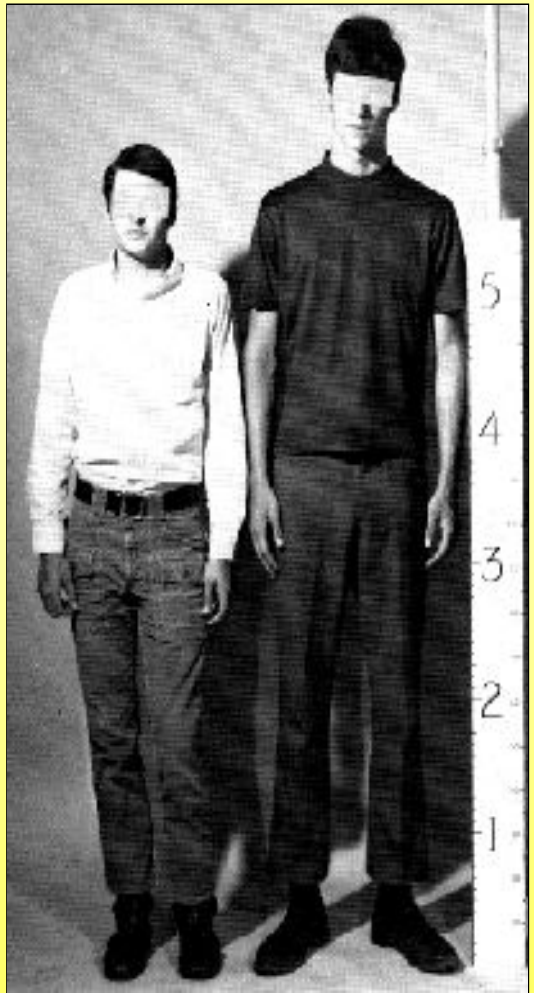
NORMAL TAURODONTISM

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

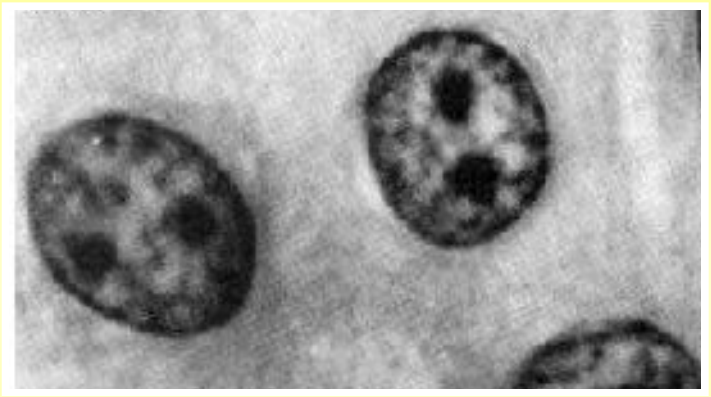


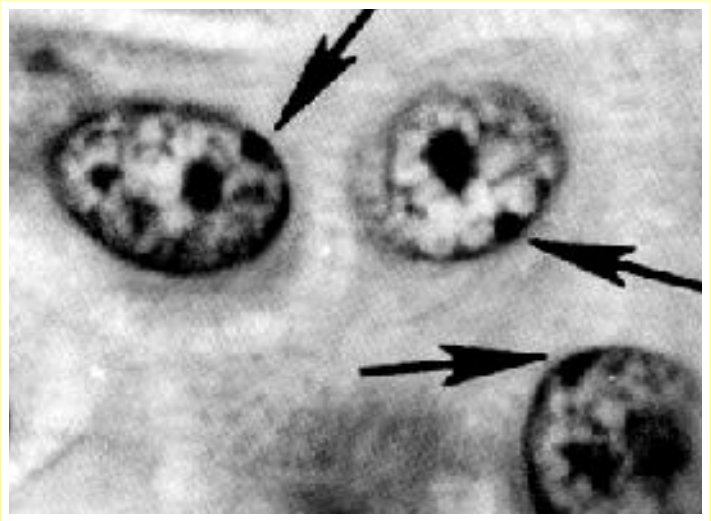
TWINS – TALLER 47,XYY

task 7, p. 81

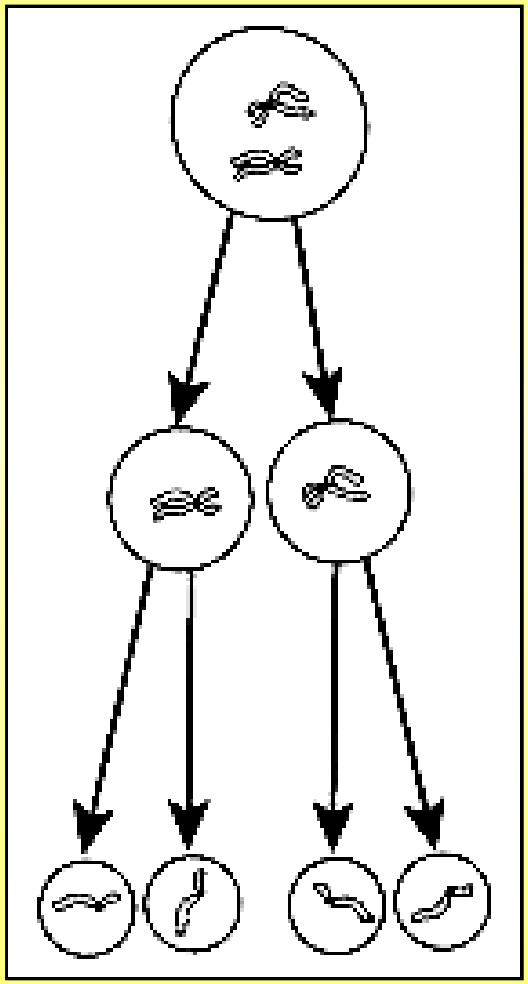
Enumerate the Barr bodies in:

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,XXXY	h) 2
i) 47,XY,+13	i) 0

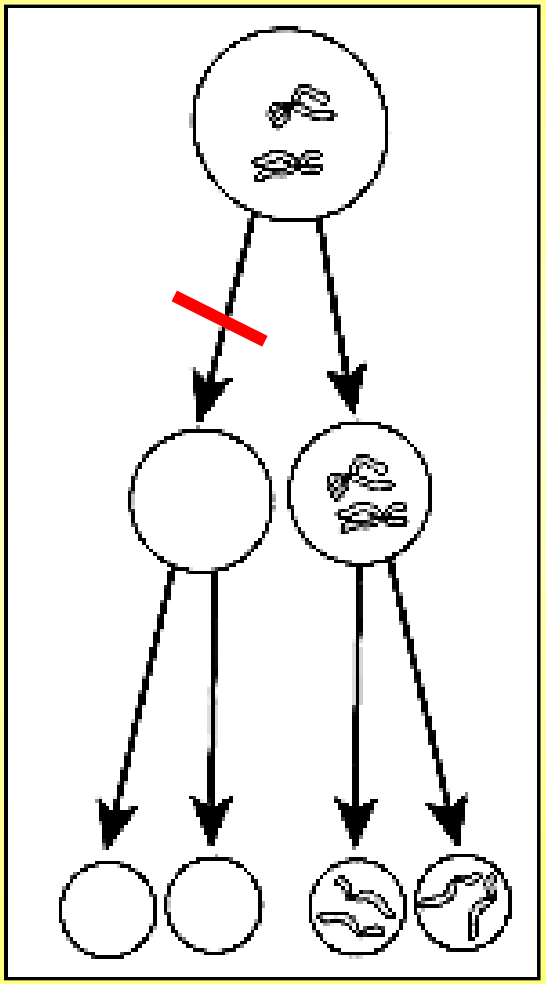




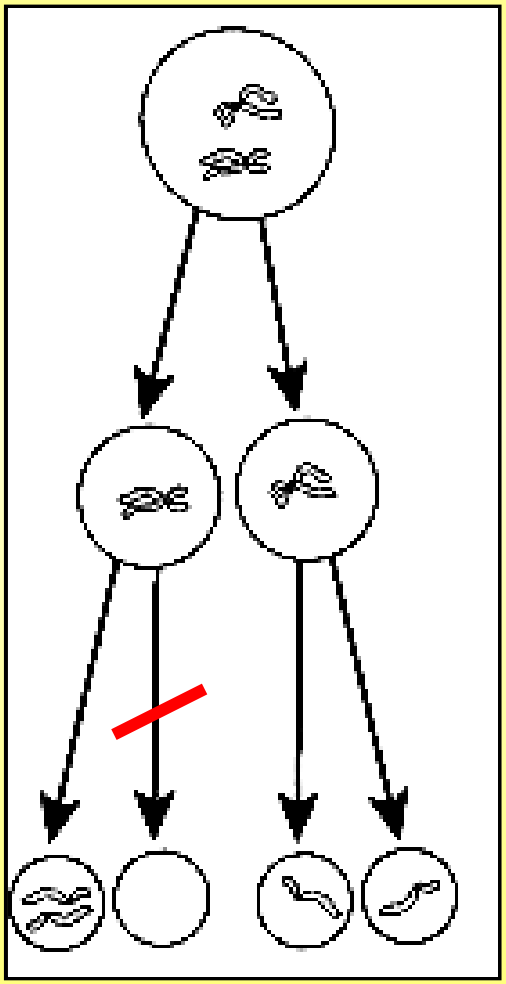
NONDISJUNCTION



NORMAL

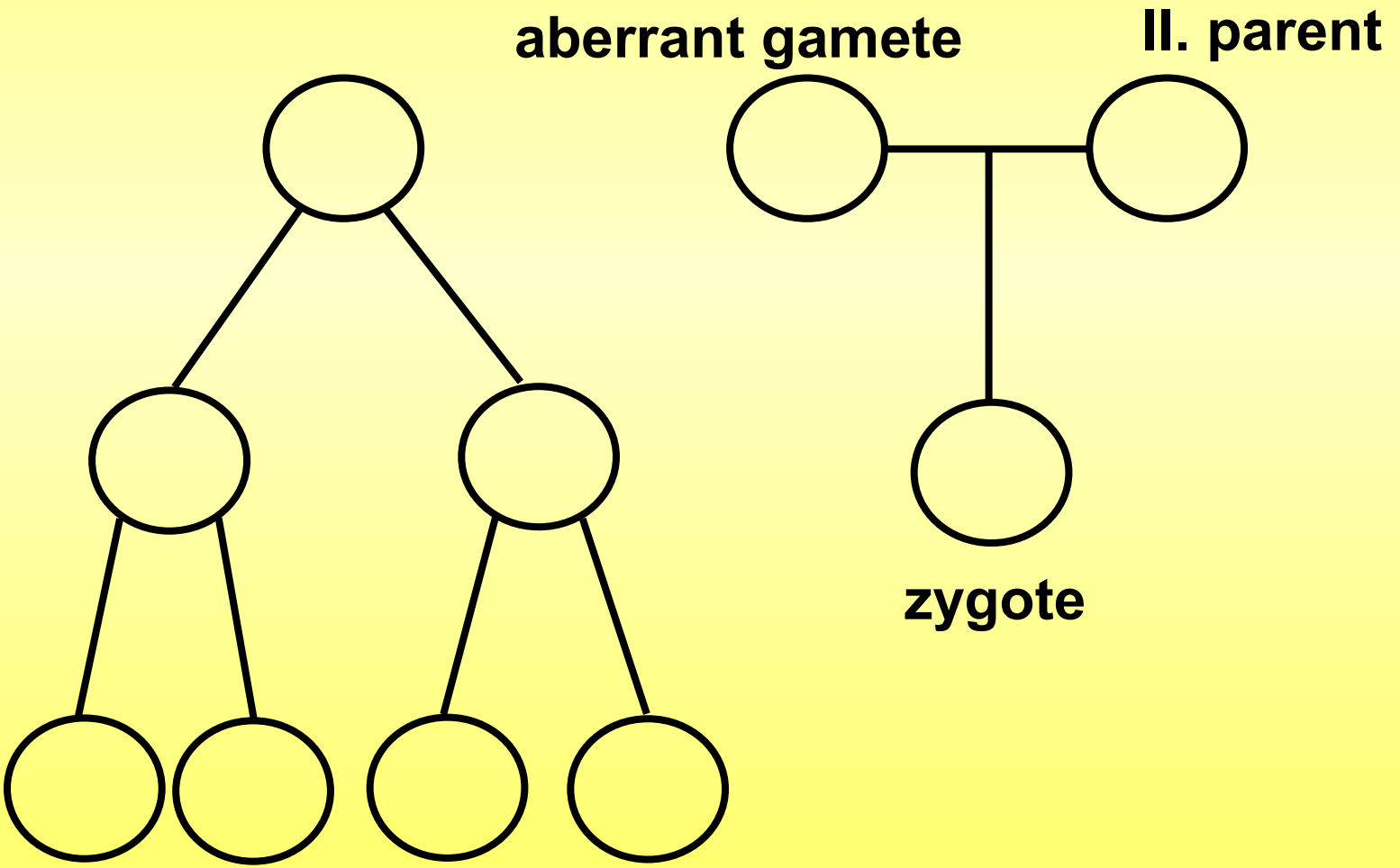


NONDIS. MI

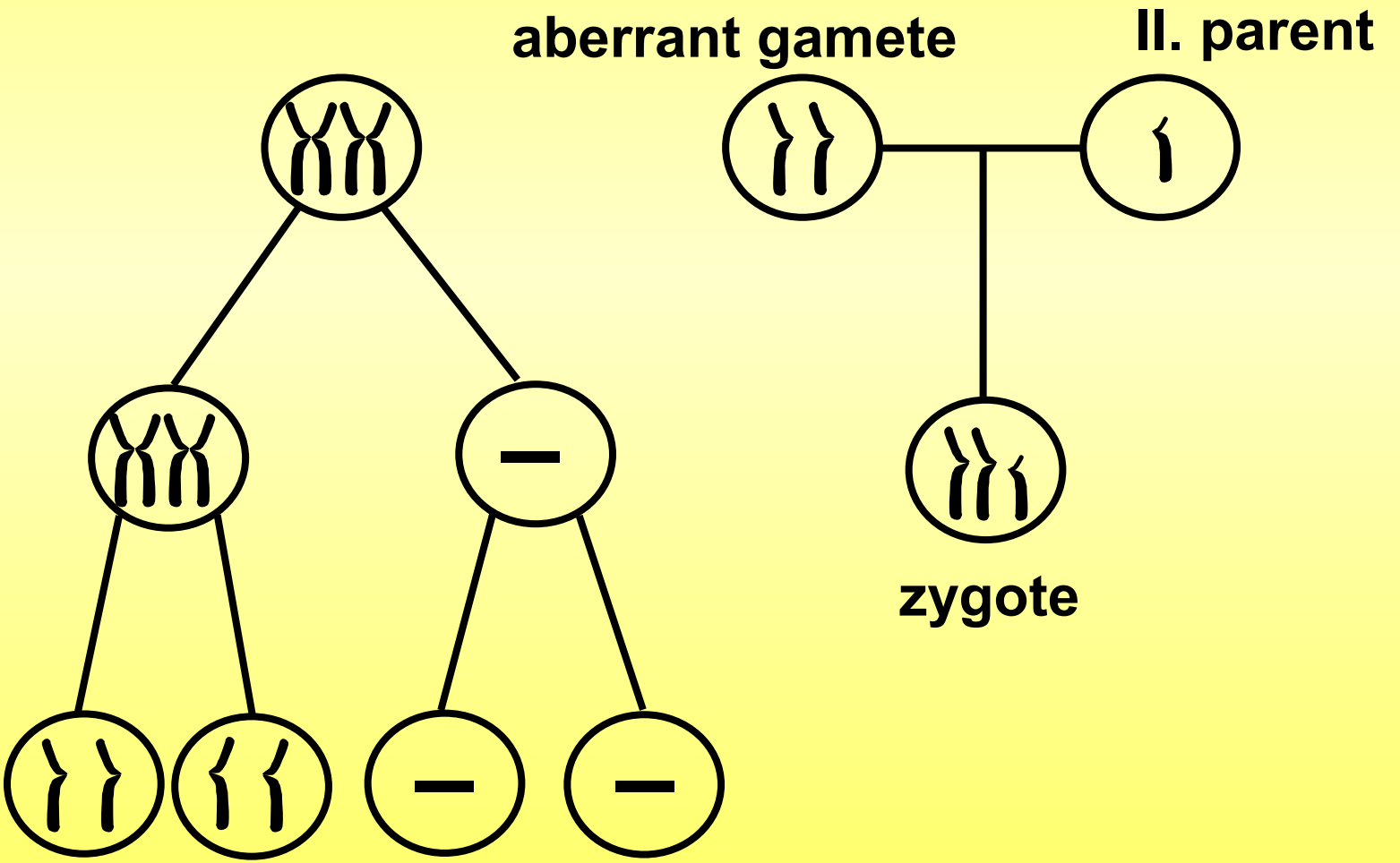


NONDIS. MII

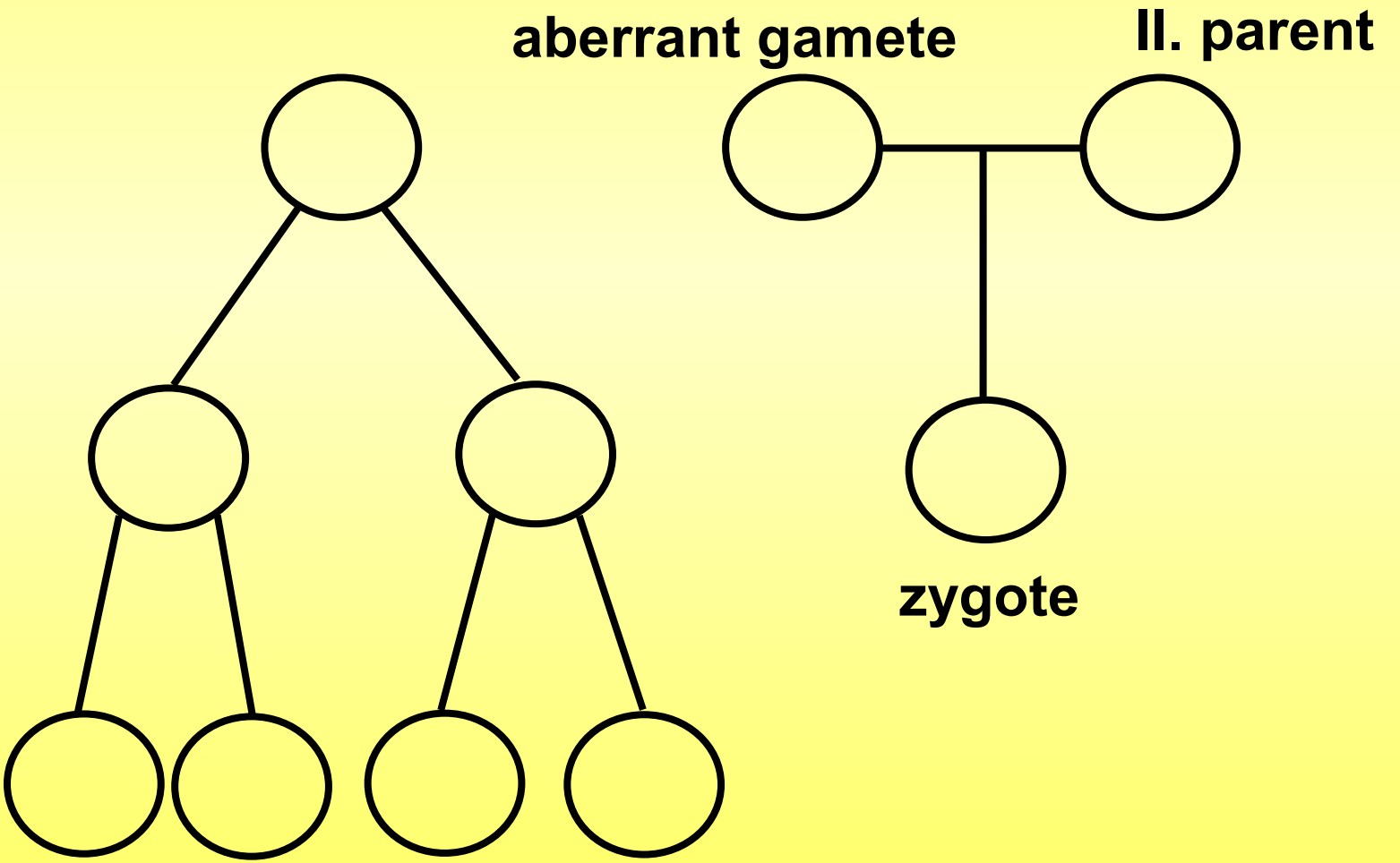
Fill in the scheme of the nondisjunction in meiosis I in mother leading to the birth of a child with Klinefelter syndrome



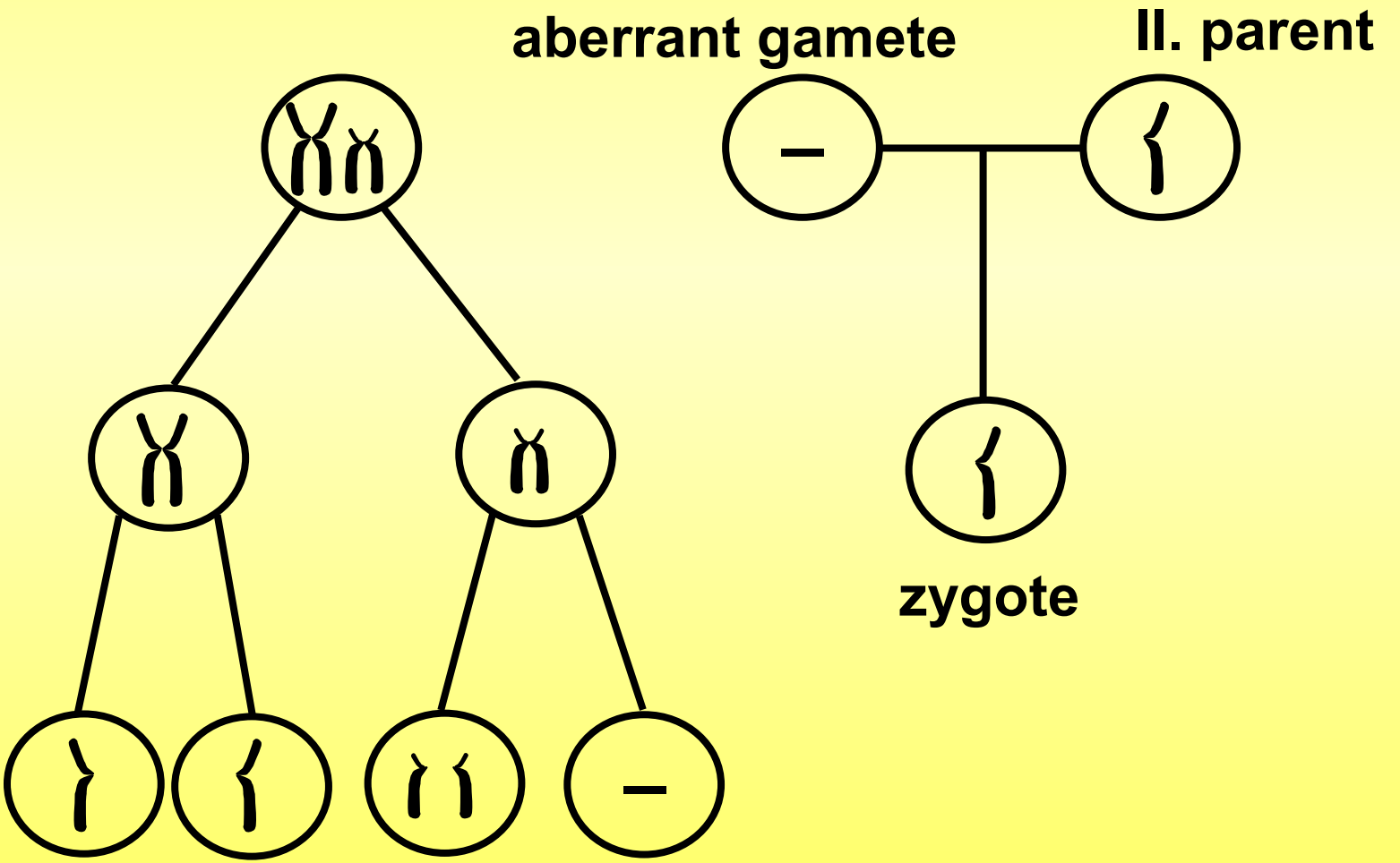
Fill in the scheme of the nondisjunction in meiosis I in mother leading to the birth of a child with Klinefelter syndrome



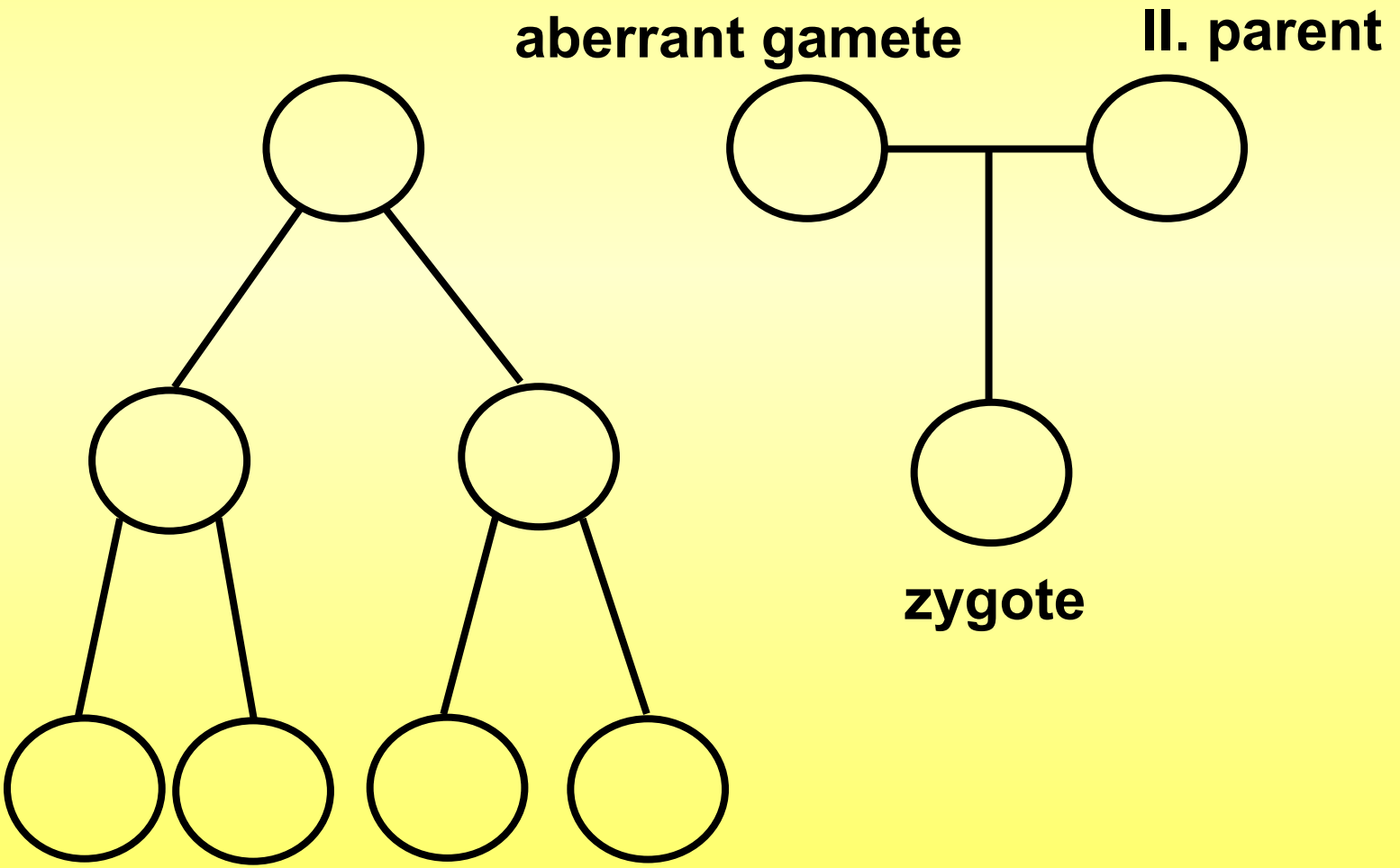
Fill in the scheme of the nondisjunction in meiosis II in father leading to the birth of a child with Turner syndrome



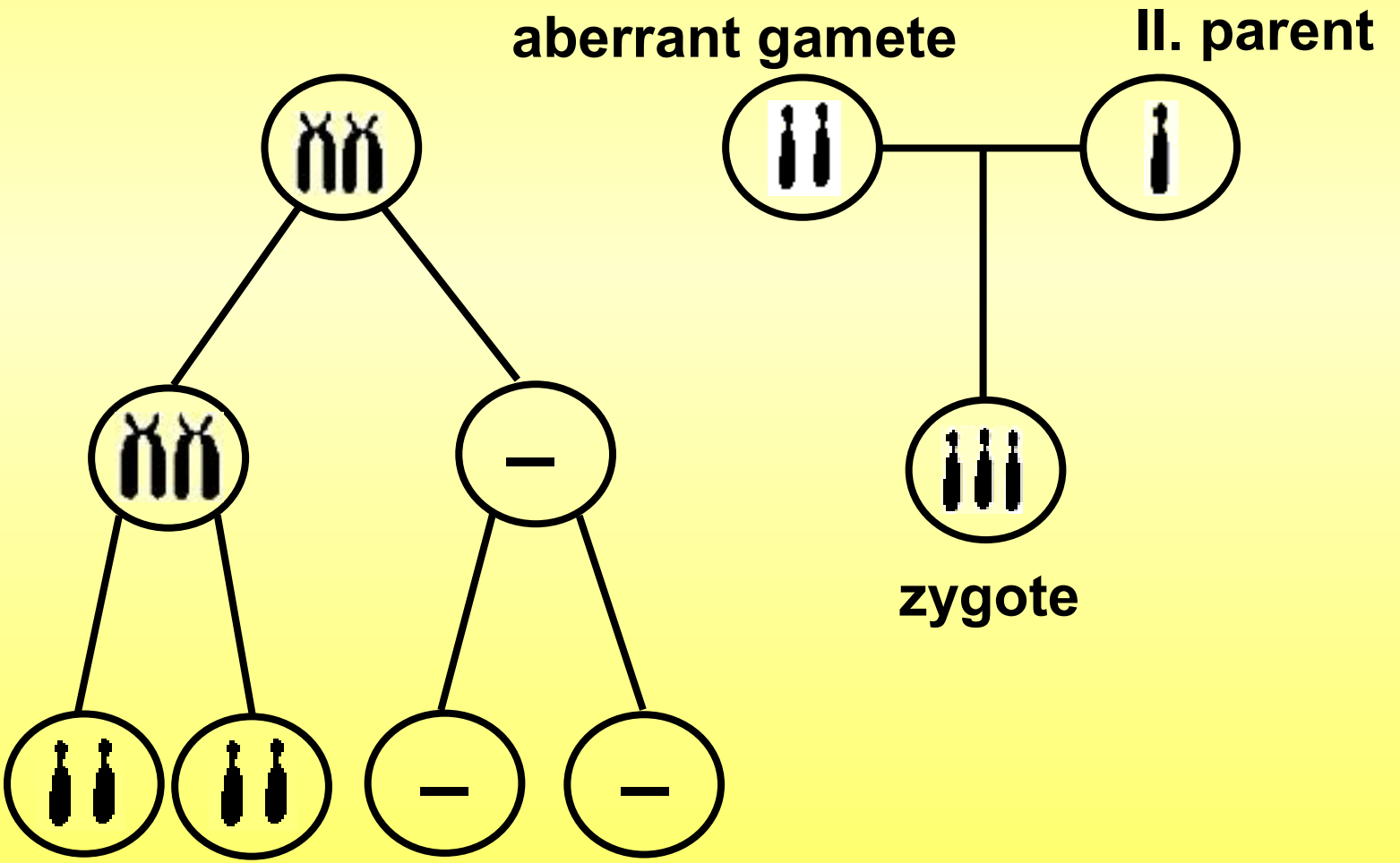
Fill in the scheme of the nondisjunction in meiosis II in father leading to the birth of a child with Turner syndrome



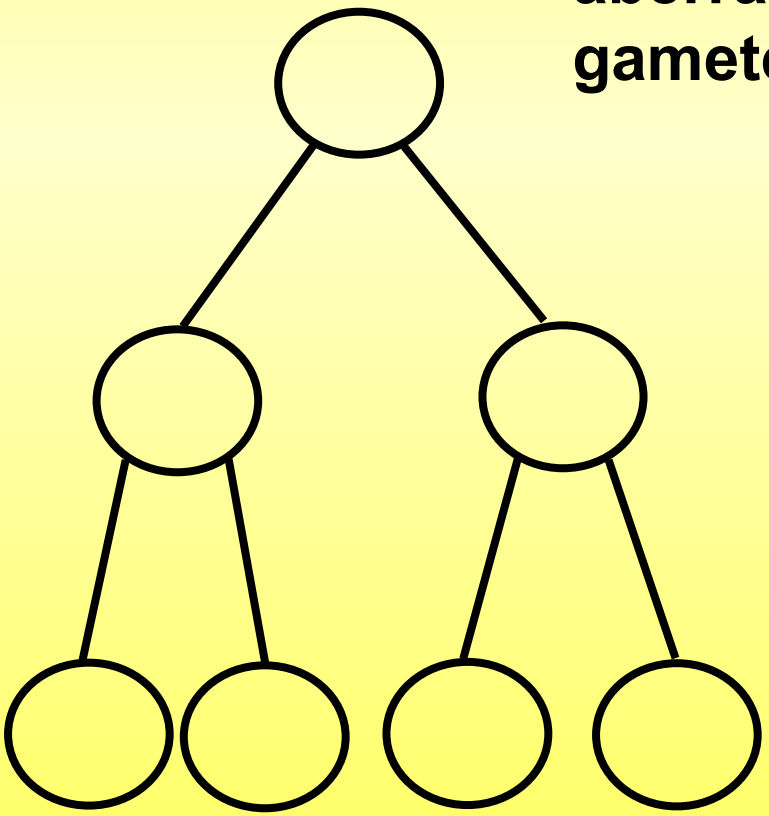
Fill in the scheme of the nondisjunction in meiosis I in one parent leading to the birth of a child with Down syndrome

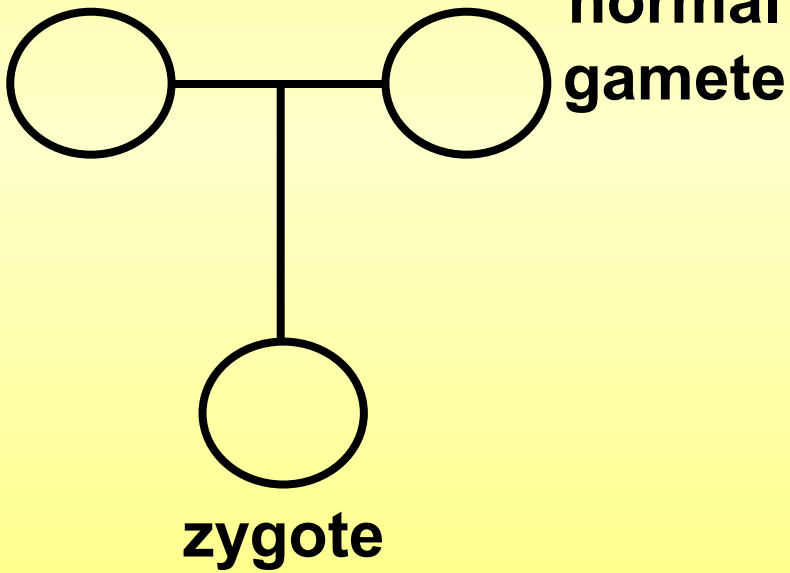


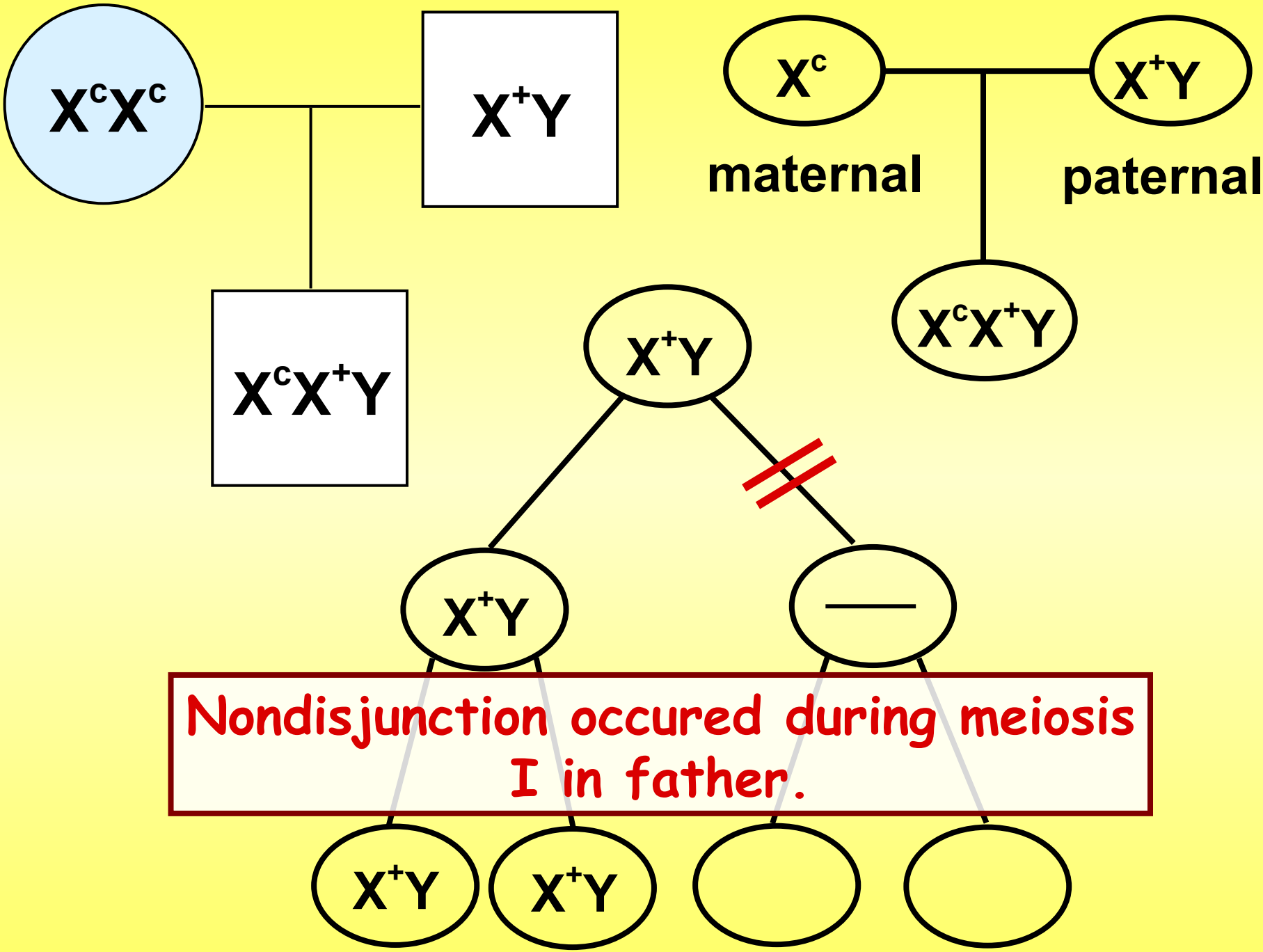
Fill in the scheme of the nondisjunction in meiosis I in one parent leading to the birth of a child with Down syndrome



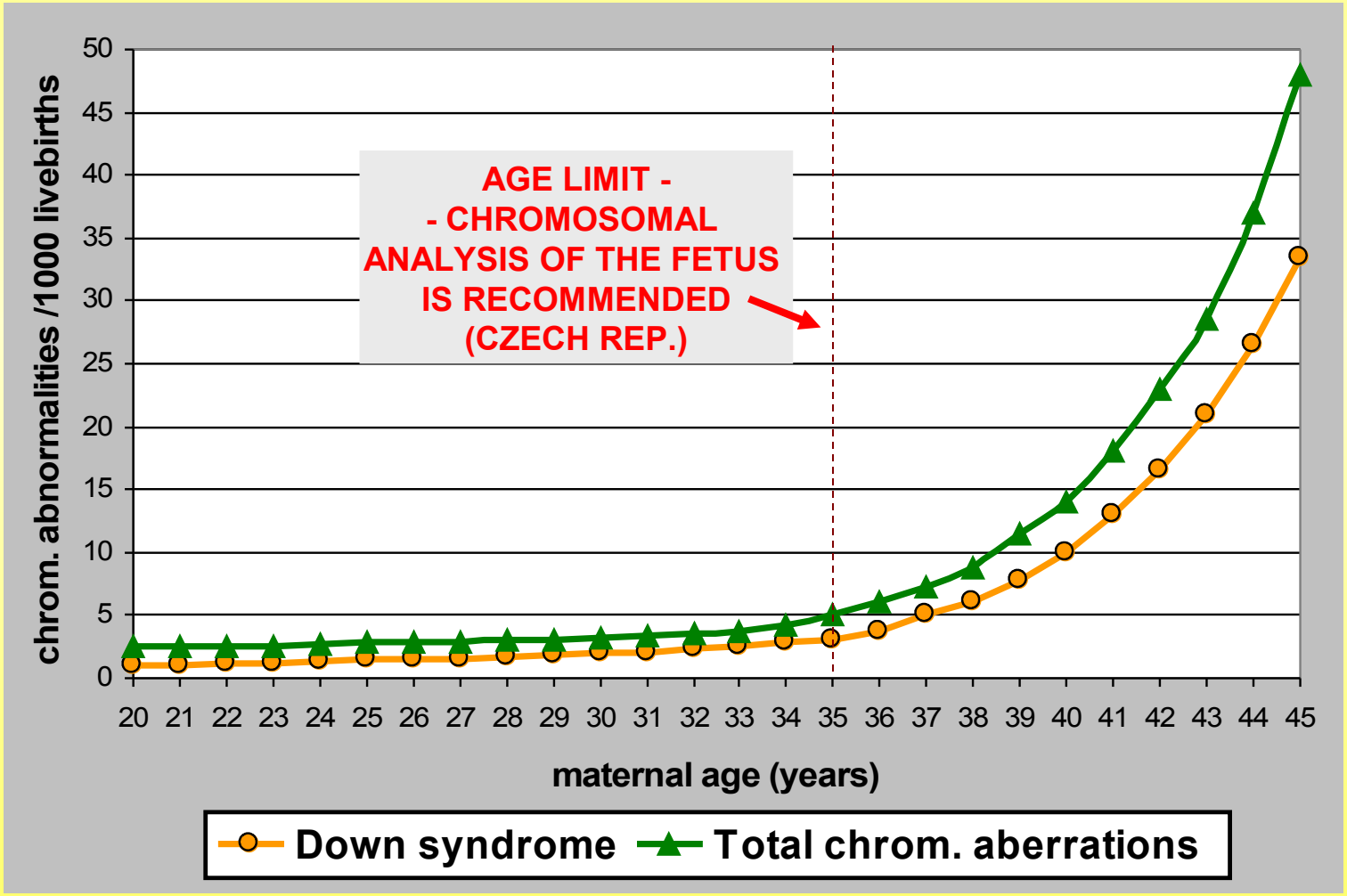
task 12, p.86: A colour-blind mother and father with normal colour vision have a son with normal vision whose karyotype is 47,XXY. Both parents have normal karyotype. In which parent and at which meiotic division did nondisjunction occur?






X^cX^c
X^+Y
X^cX^+Y
X^c
X^+Y
maternal
paternal
X^cX^+Y
X^+Y
X^+Y
—
Nondisjunction occured during meiosis I in father.
X^+Y
X^+Y

INCREASING FREQUENCY OF THE CHROMOSOMAL ABERRATIONS WITH THE AGE OF THE MOTHER

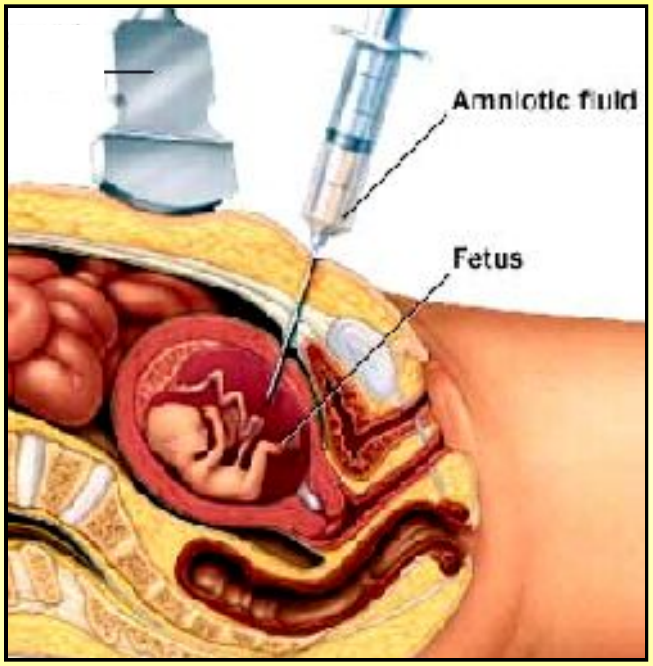


PRENATAL DIAGNOSTICS

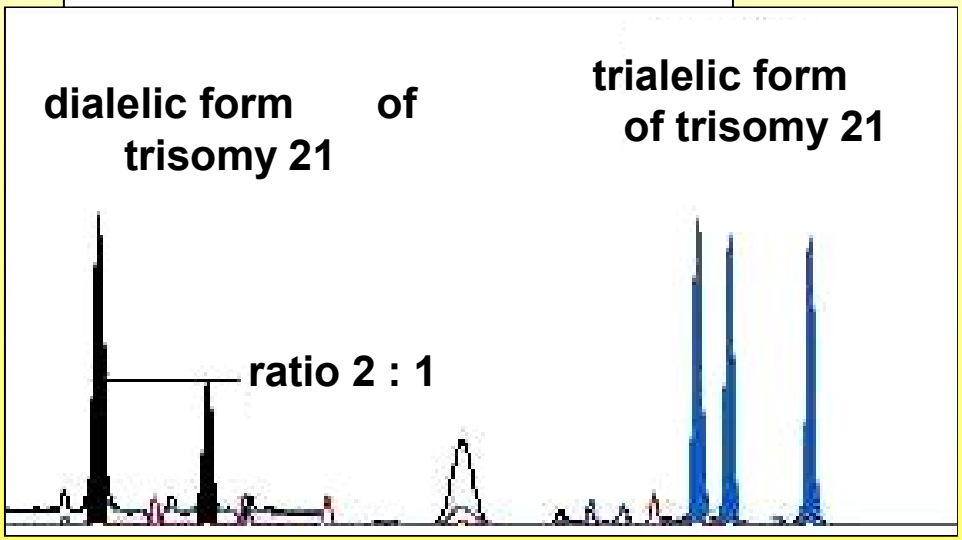
CHORIONIC VILLI SAMPLE

AMNIOCENTESIS (16.-18.w.)

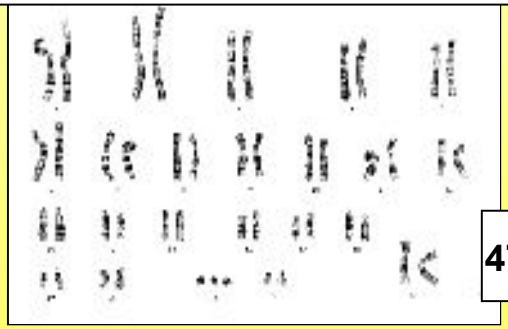
CORDOCENTESIS



amnioPCR (within 24 hours)

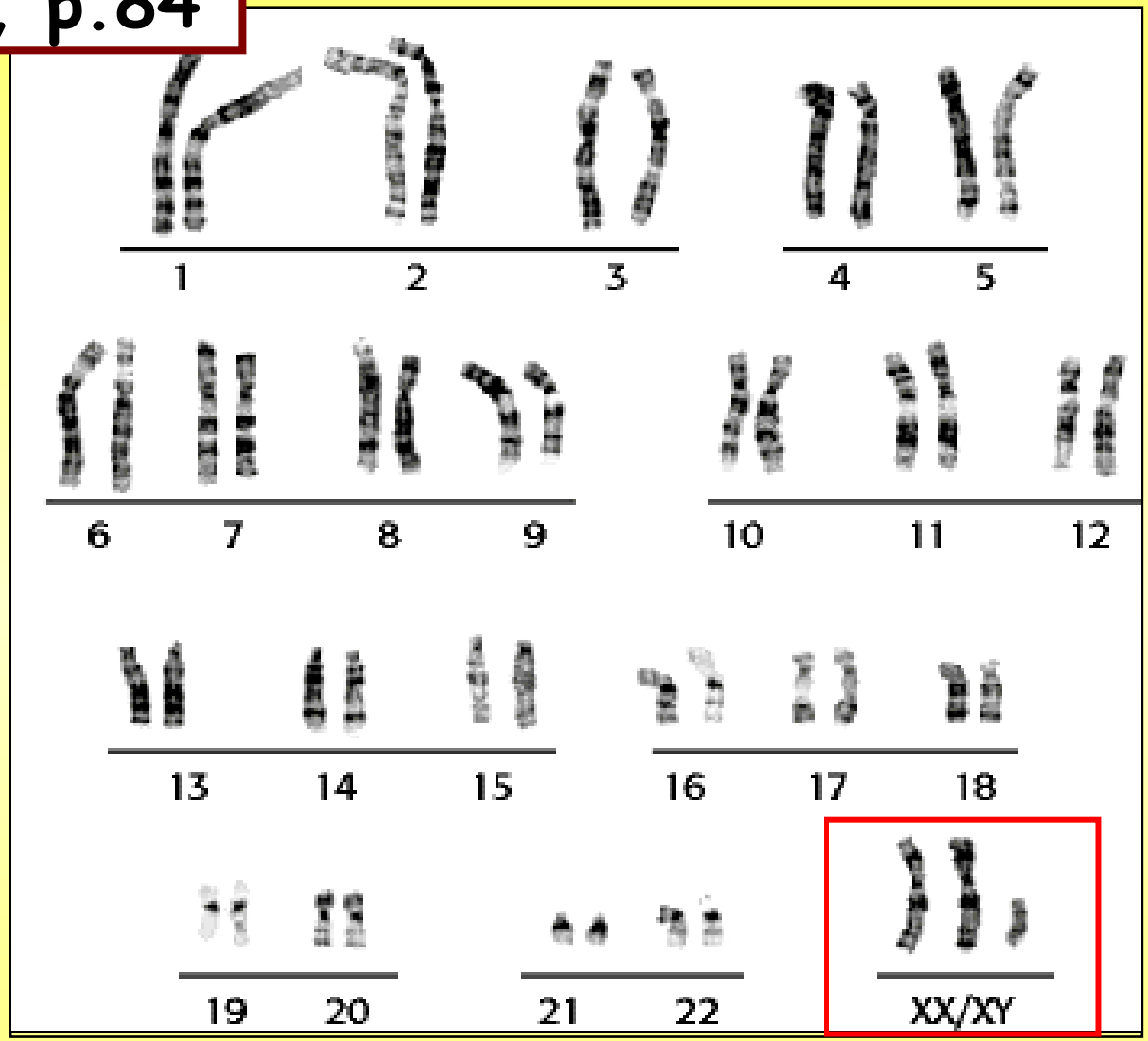


KARYOTYPE (cca 2 wks)



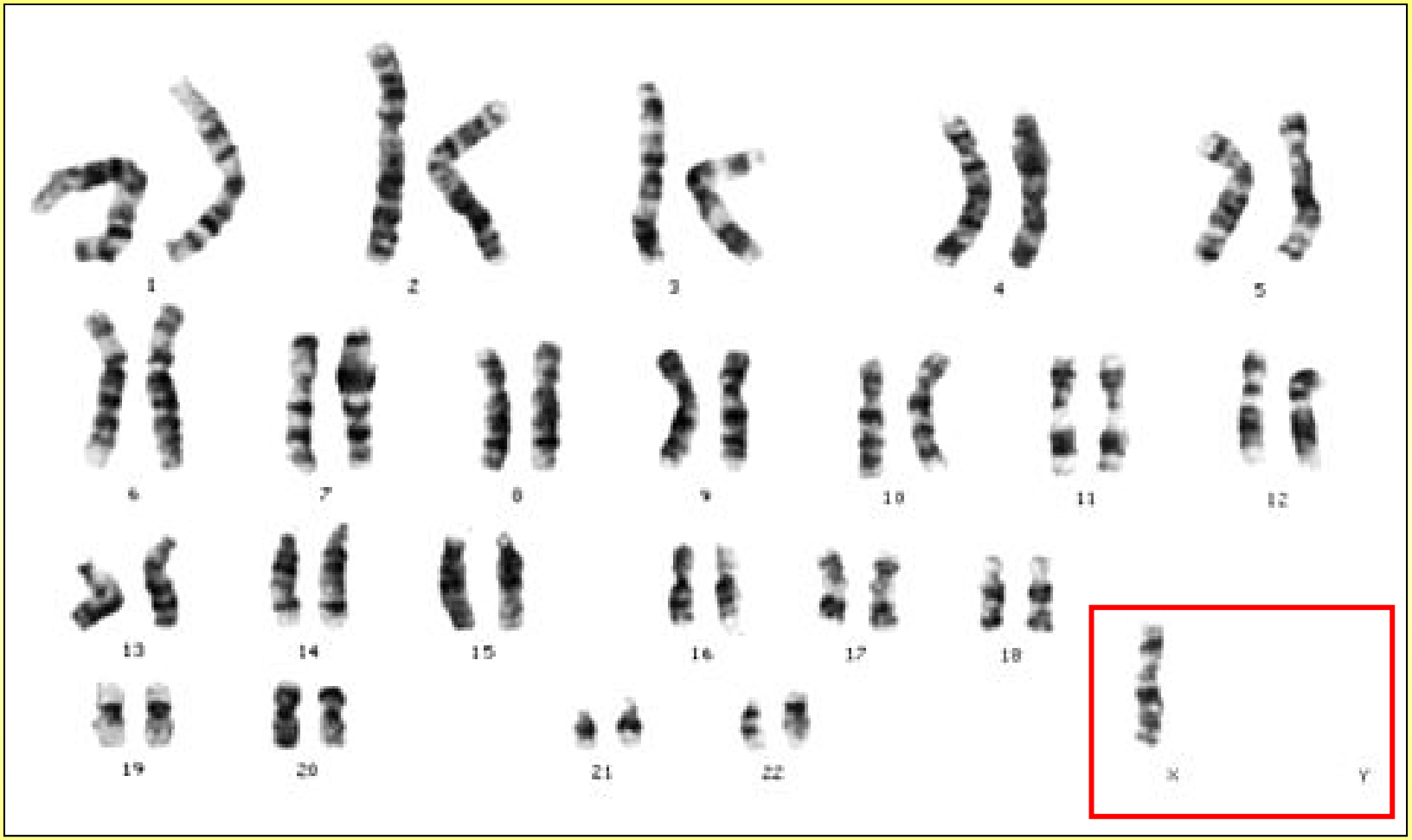
47,XX,+21

task 10, p.84



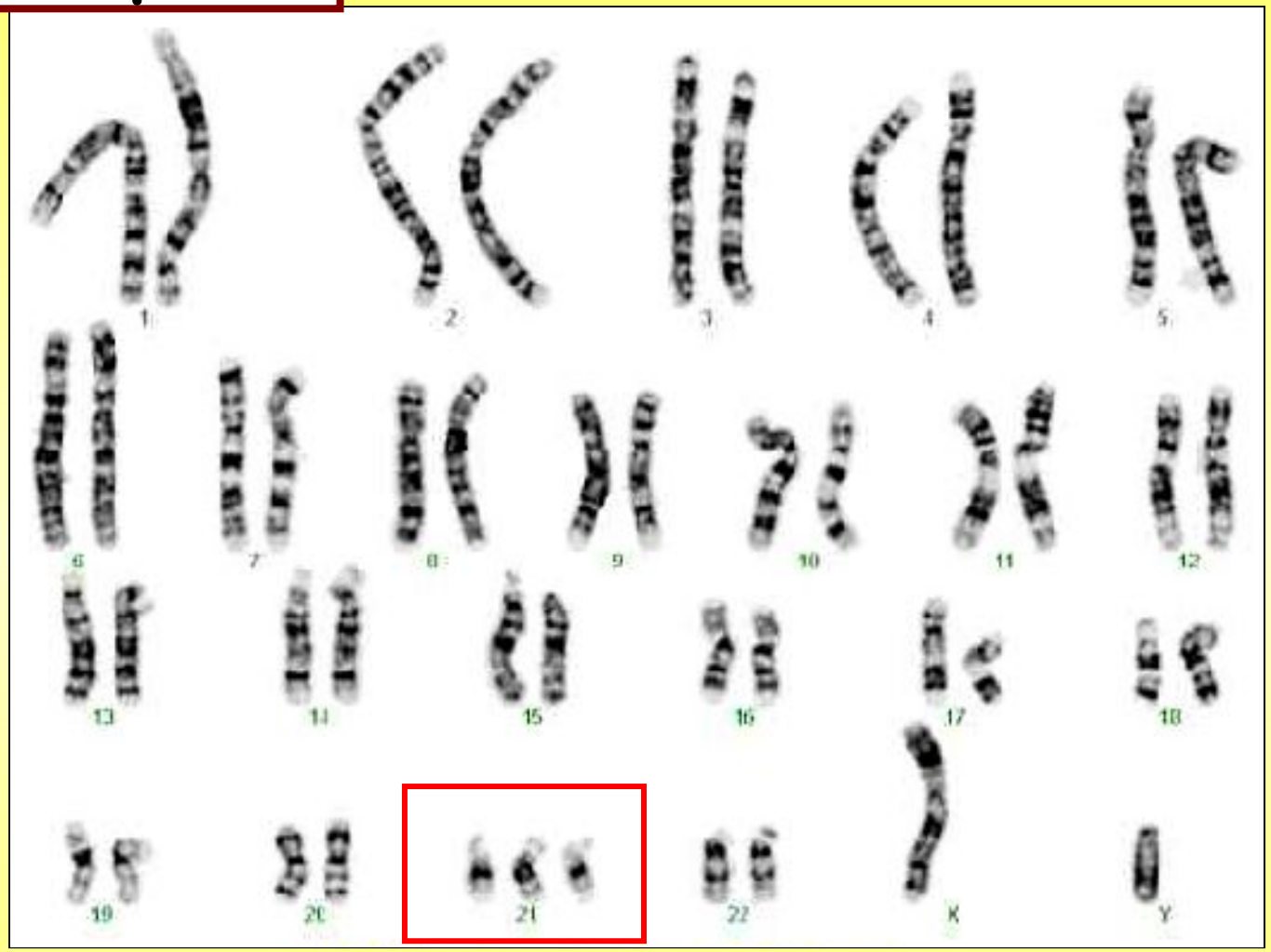
47,XXY

task 9, p. 82

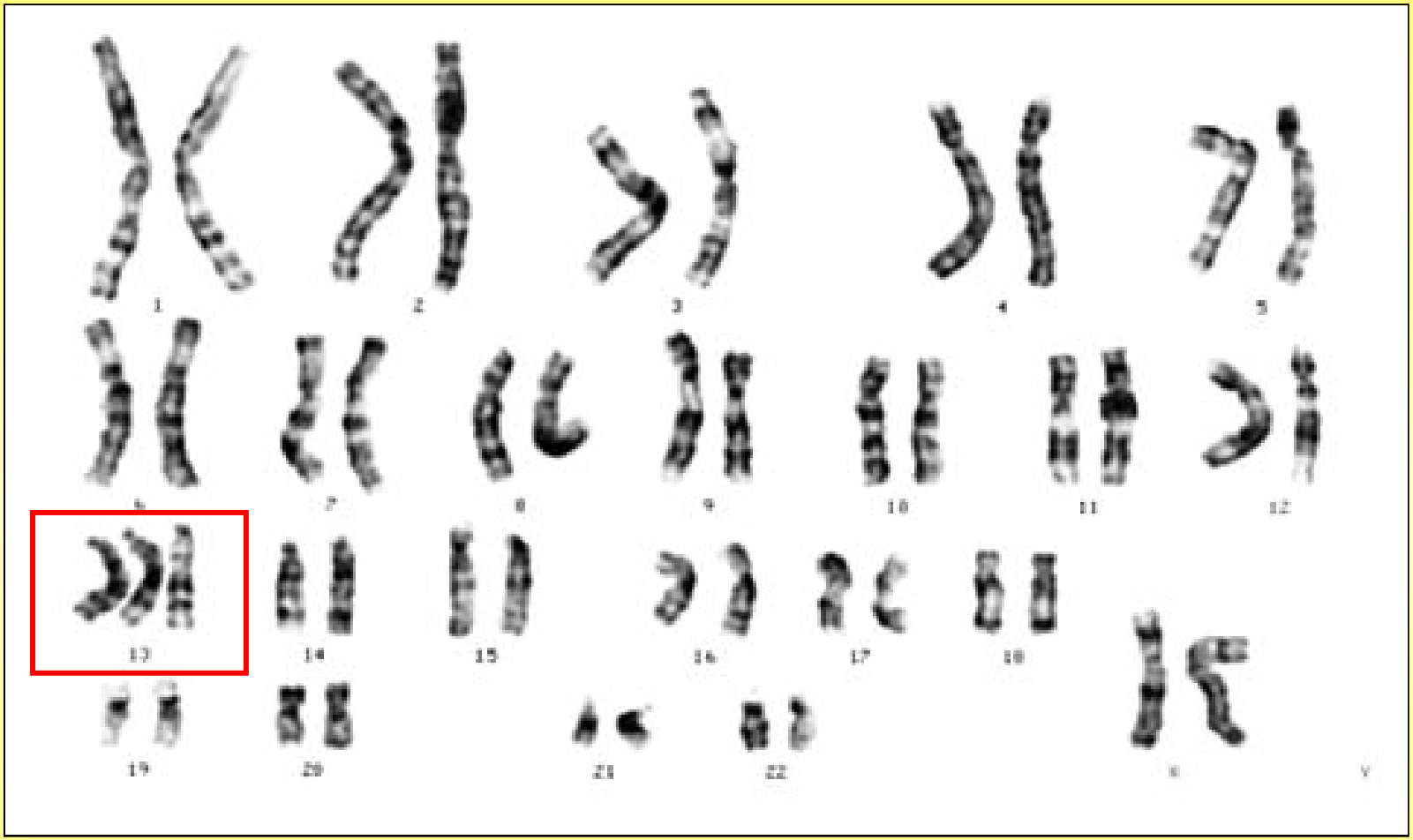


45,X

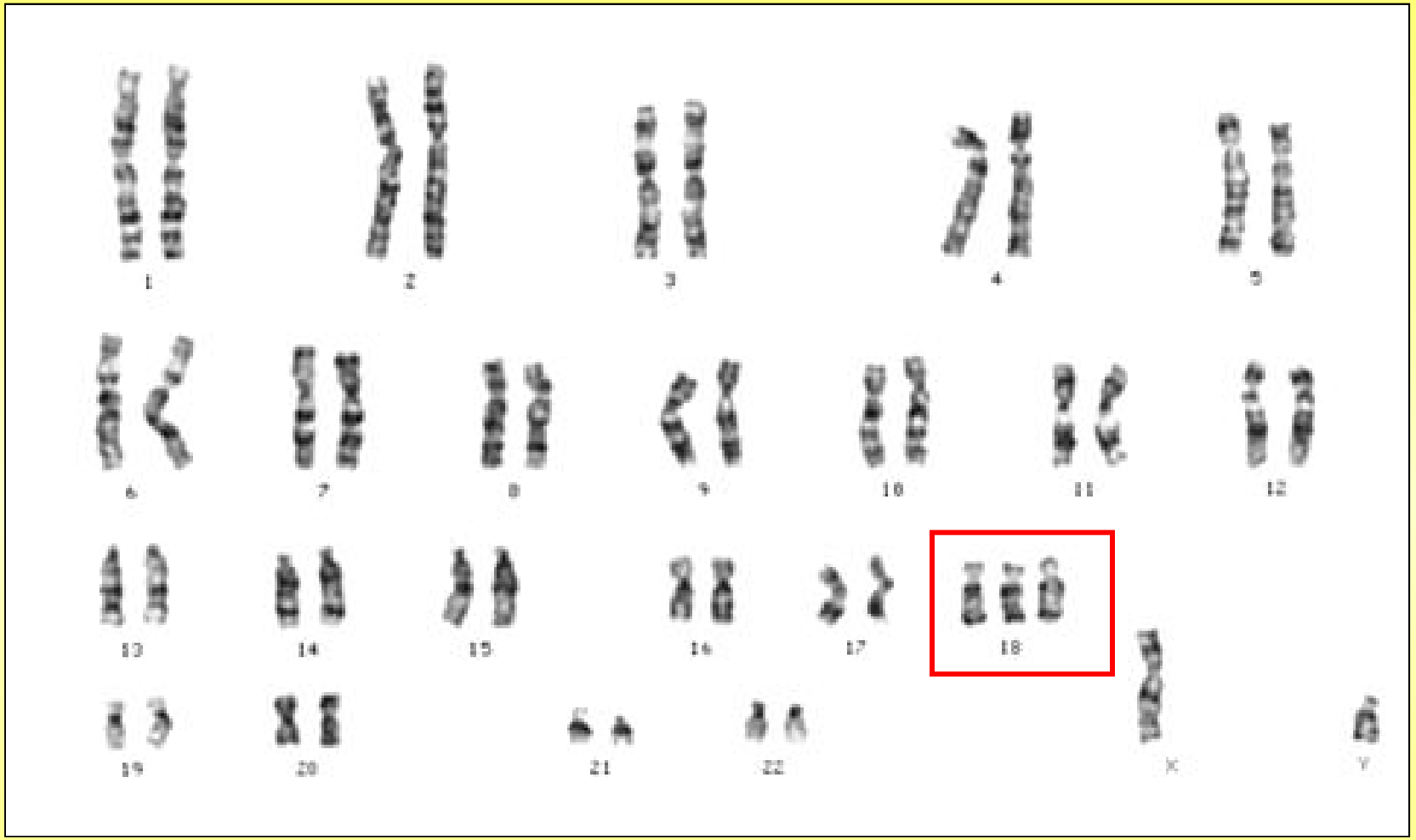
task 11, p. 86



47,XY,+21



47,XX,+13



47,XY,+18