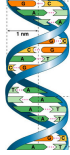


AP Biology

Chapter 15.

Chromosomal Abnormalities



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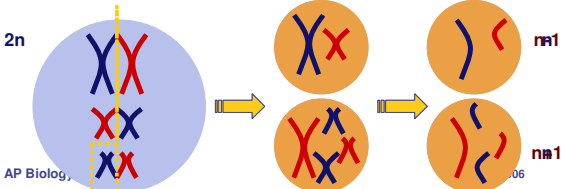
Chromosomal abnormalities

- Incorrect number of chromosomes
 - nondisjunction**
 - chromosomes don't separate properly during meiosis
 - breakage of chromosomes
 - deletion
 - duplication
 - inversion
 - translocation

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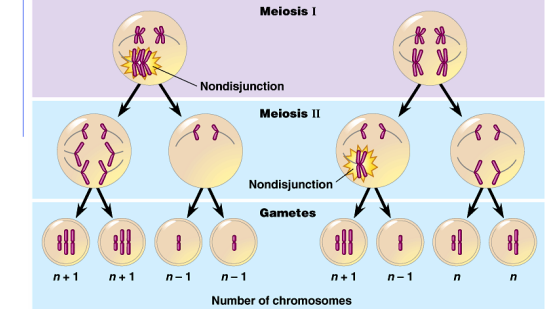
Nondisjunction

- Problems with meiotic spindle cause errors in daughter cells
 - tetrad chromosomes do not separate properly during Meiosis 1
 - sister chromatids fail to separate during Meiosis 2
 - too many or too few chromosomes



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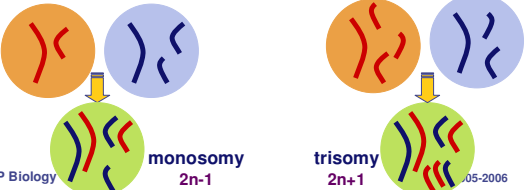
Alteration of chromosome number



AP | (a) Nondisjunction of homologous chromosomes in meiosis I (b) Nondisjunction of sister chromatids in meiosis II

Nondisjunction

- Baby has wrong chromosome number
 - trisomy**
 - cells have 3 copies of a chromosome
 - monosomy**
 - cells have only 1 copy of a chromosome



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Human chromosome disorders


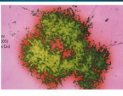

- High frequency in humans
 - most embryos are spontaneously aborted
 - alterations are too disastrous
 - developmental problems result from biochemical imbalance
- Certain conditions are tolerated
 - upset the balance less = **survive**
 - characteristic set of symptoms = **syndrome**

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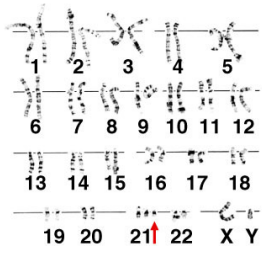

Down syndrome

- Trisomy 21
 - 3 copies of chromosome 21
 - 1 in 700 children born in U.S.
- Chromosome 21 is the smallest human chromosome
 - but still severe effects
- Frequency of Down syndrome correlates with the age of the mother

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Trisomy 21

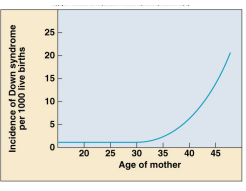



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Down syndrome & age of mother

Mother's age	Incidence of Down Syndrome
Under 30	<1 in 1000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12

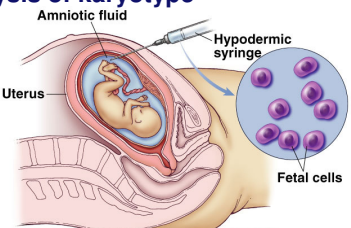


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Genetic testing

- Amniocentesis in 2nd trimester
 - sample of embryo cells
 - stain & photograph chromosomes
- Analysis of karyotype

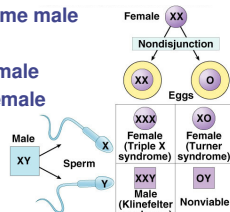


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Sex chromosomes


- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct conditions in humans
 - XXY = Klinefelter's syndrome male
 - XXX = Trisomy X female
 - XYY = Jacob's syndrome male
 - XO = Turner syndrome female



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Klinefelter's syndrome

- XXY male
 - one in every 2000 live births
 - have male sex organs, but are sterile
 - feminine characteristics
 - tall
 - normal intelligence



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Klinefelter's syndrome

AP Biology Karyotype: 47, XXY 2005-2006

Klinefelter's syndrome

XXXXY, Klinefelter's Syndrome

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Jacob's syndrome male

- **XYY Males**
 - ◆ 1 in 1000 live male births
 - ◆ extra Y chromosome
 - ◆ somewhat taller than average
 - ◆ more active
 - ◆ slight learning disabilities
 - ◆ delayed emotional immaturity
 - ◆ normal intelligence, normal sexual development

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XYY Males

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Trisomy X

- **XXX**
 - ◆ 1 in every 2000 live births
 - ◆ produces healthy females
 - Why?

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Turner syndrome

- **Monosomy X or XO**
 - ◆ 1 in every 5000 births
 - ◆ varied degree of effects
 - ◆ webbed neck
 - ◆ short stature
 - ◆ immature sterile females

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Turner syndrome

Short stature

Low hairline

Shield-shaped thorax

Widely spaced nipples

Shortened metacarpal IV

Small finger nails

Brown spots (nevi)

Characteristic facial features

Fold of skin

Constriction of aorta

Poor breast development

Elbow deformity

Rudimentary ovaries

Gonadal streak (underdeveloped gonadal structures)

No menstruation

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Changes in chromosome structure

(a) A **deletion** removes a chromosomal segment.

(b) A **duplication** repeats a segment.

(c) An **inversion** reverses a segment within a chromosome.

(d) A **translocation** moves a segment from one chromosome to another, non-homologous one.

Reciprocal translocation

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Any Questions??

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