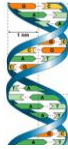


AP Biology

Chapter 15.

Chromosomal Abnormalities



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Chromosomal abnormalities

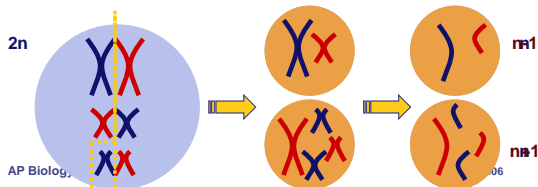
- **Incorrect number of chromosomes**
 - ◆ **nondisjunction**
 - chromosomes don't separate properly during meiosis
- ◆ **breakage of chromosomes**
 - deletion
 - duplication
 - inversion
 - translocation

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Nondisjunction

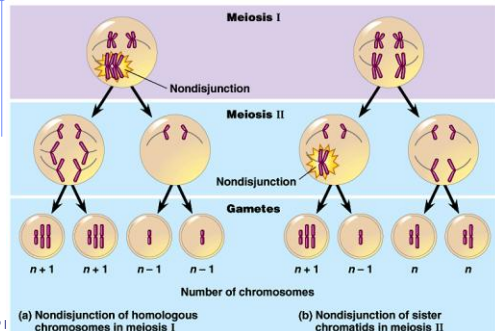
- **Problems with meiotic spindle cause errors in daughter cells**
 - ◆ tetrad chromosomes do not separate properly during Meiosis 1
 - ◆ sister chromatids fail to separate during Meiosis 2
 - ◆ too many or too few chromosomes



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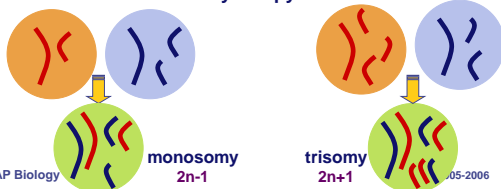
Alteration of chromosome number



AP I

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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AP Biology

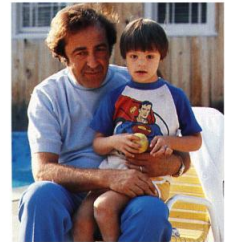
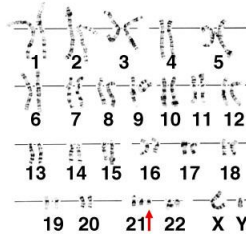
Down syndrome

- 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

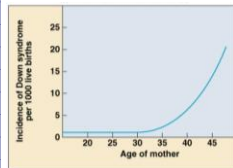


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Down syndrome & age of mother

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



AP B

2005-2006

Genetic testing

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

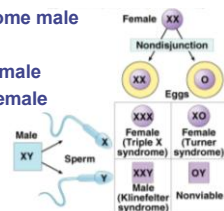


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Sex chromosomes

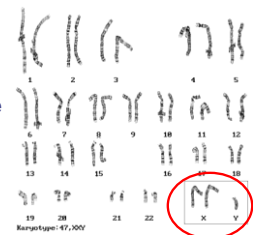
- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct conditions in humans
 - XXY = Klinefelter's syndrome male
 - XXX = Trisomy X female
 - XYY = Jacob's syndrome male
 - XO = Turner syndrome female



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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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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

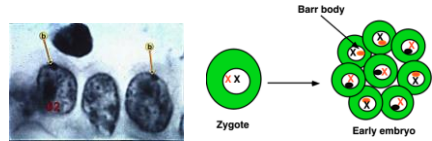


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Trisomy X

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

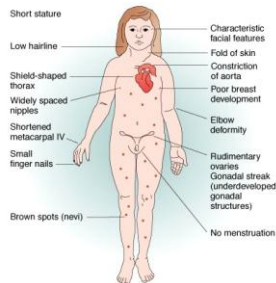


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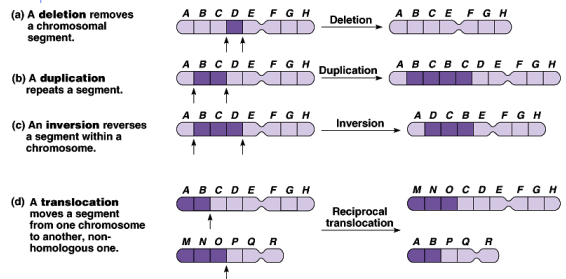
Turner syndrome

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



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Changes in chromosome structure



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Any Questions??

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