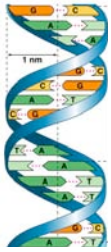
Chapter 15.



Chromosomal Abnormalities

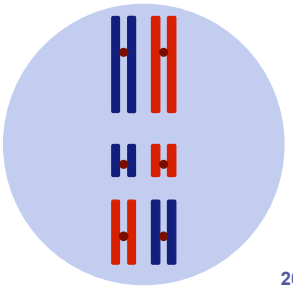
AP Biology 2004-2005

Chromosomal abnormalities

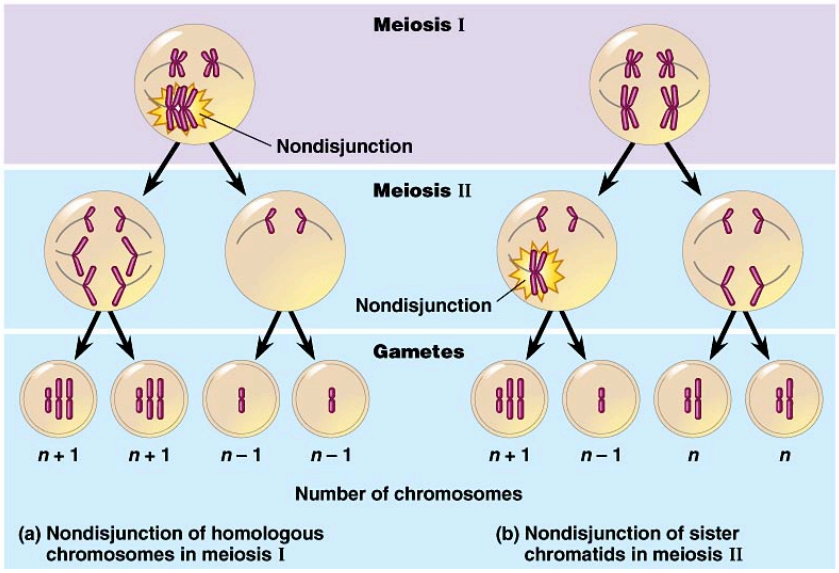
- Incorrect number of chromosomes
  - nondisjunction
    - chromosomes don't separate properly during meiosis
  - breakage of chromosomes
    - deletion
    - duplication
    - inversion
    - translocation

Nondisjunction

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



Alteration of chromosome number



AP

Nondisjunction

- Baby will have wrong chromosome number
  - Trisomy
    - cells have 3 copies of a chromosome
  - Monosomy
    - cells have only 1 copy of a chromosome

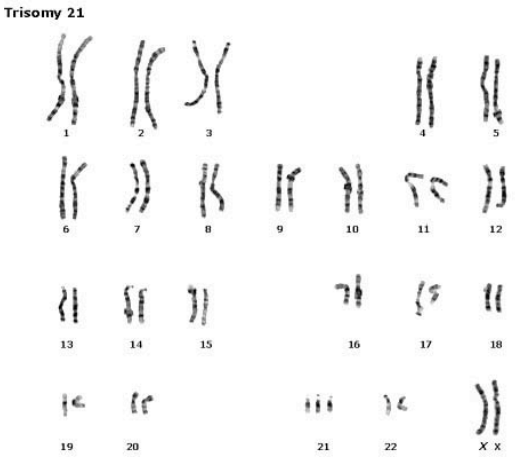
Human chromosome disorders

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

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

Trisomy 21


Trisomy 21
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X X


Benjamin Cummings

Down syndrome & age of mother

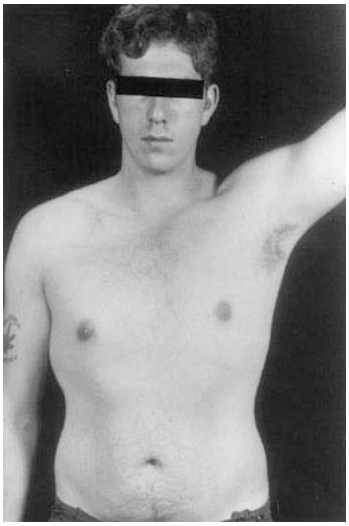
Mother's age	Incidence of Down Syndrome
Under 30	<1 in 1000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12

Sex chromosomes

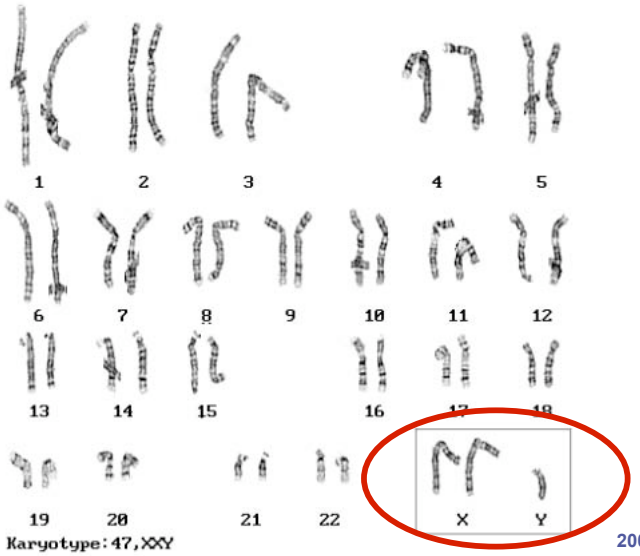
- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct conditions in humans
  - XXY = Klinefelter's syndrome male
  - XXX = Trisomy X female
  - XYY = Jacob's syndrome male
  - XO = Turner syndrome female

Klinefelter's syndrome

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



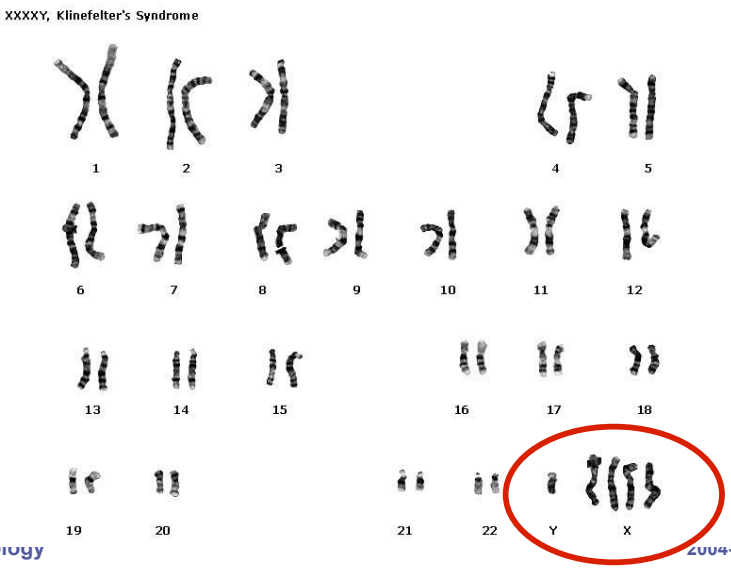
Klinefelter's syndrome



AP Biology Karyotype: 47,XXY 2004-2005

How many Barr bodies would you expect?

Klinefelter's syndrome



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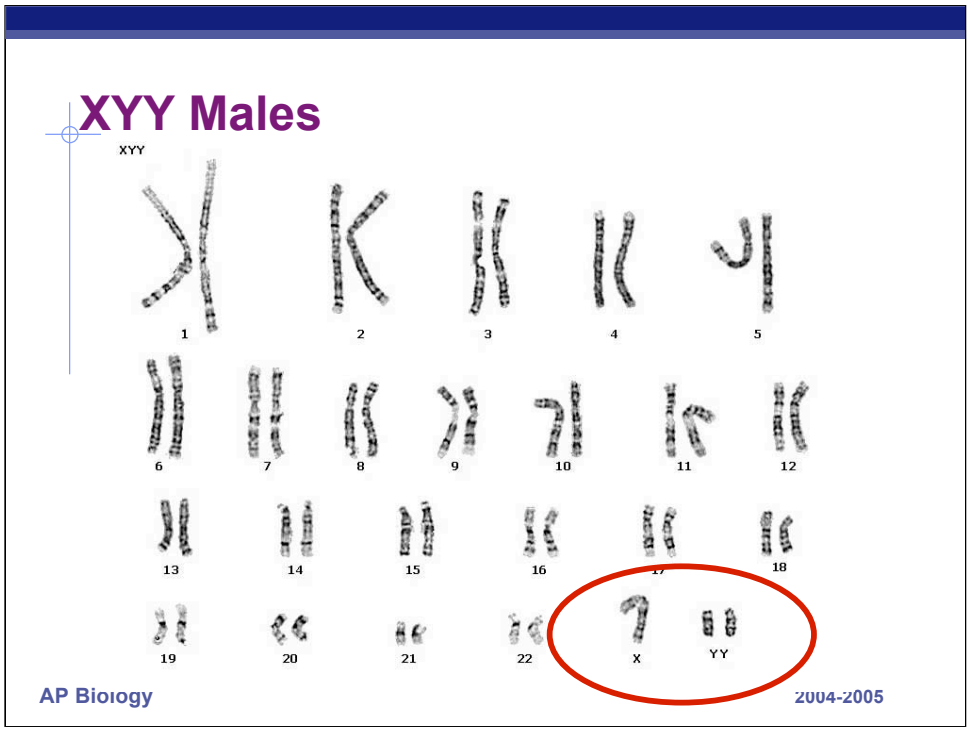
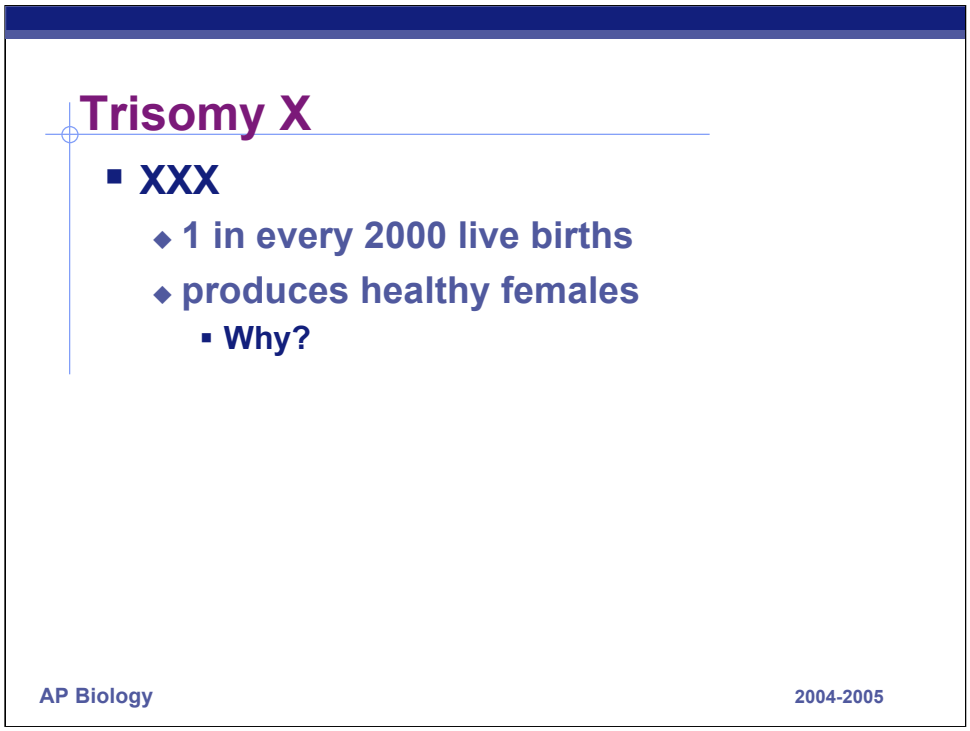
How many Barr bodies would you expect?

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



How many Barr bodies would you expect?



How many Barr bodies would you expect?

Turner syndrome

- **Monosomy X or X0**
  - 1 in every 5000 births
  - varied degree of effects
  - webbed neck
  - short stature
  - immature sterile females

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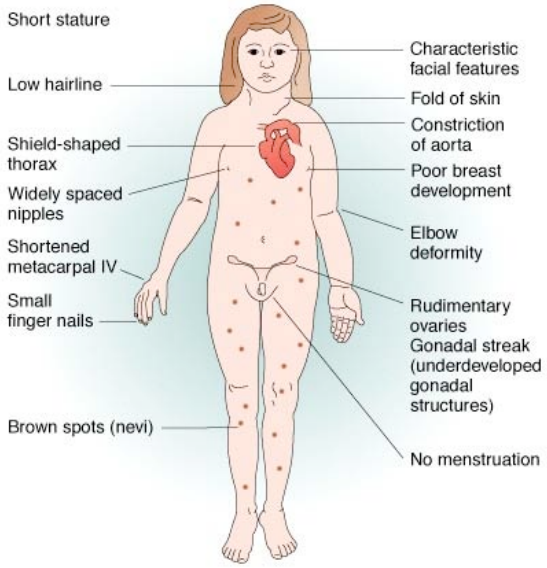
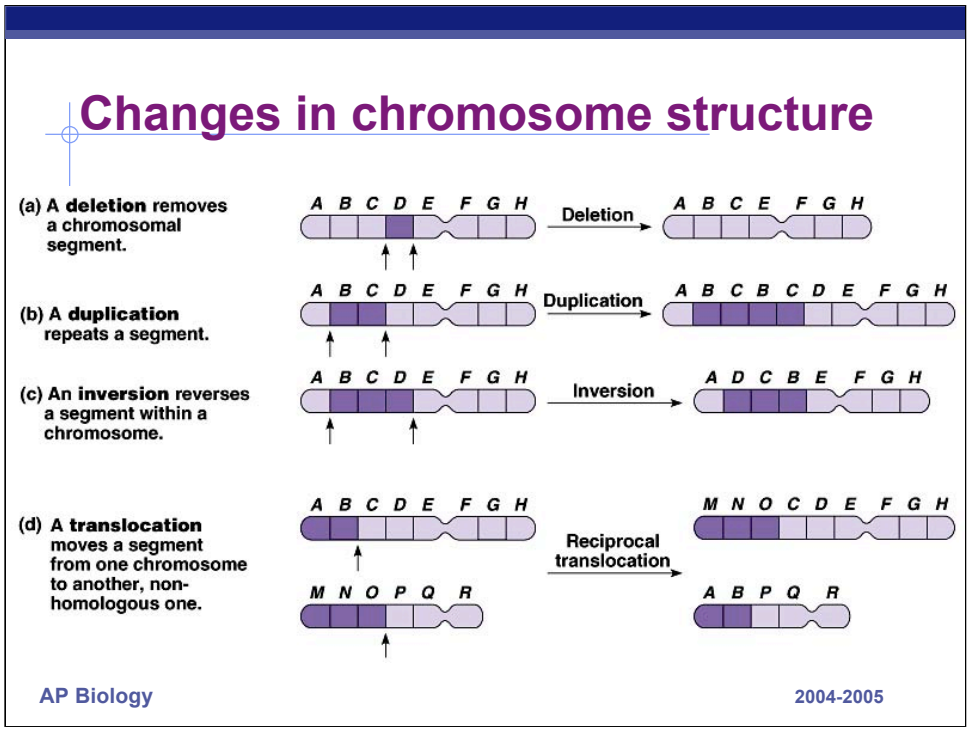
How many Barr bodies would you expect?

Turner syndrome





During what process would his happen?

Genetic testing

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