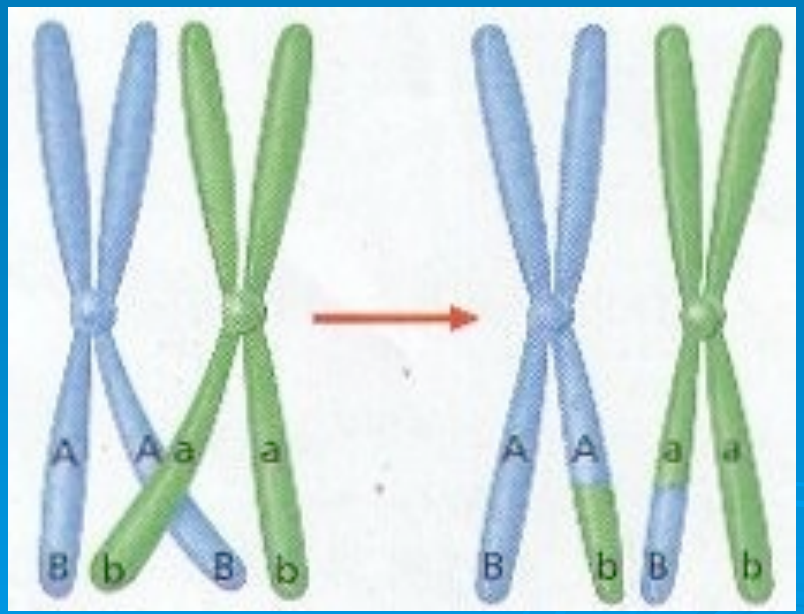
Meiosis & Karyotyping

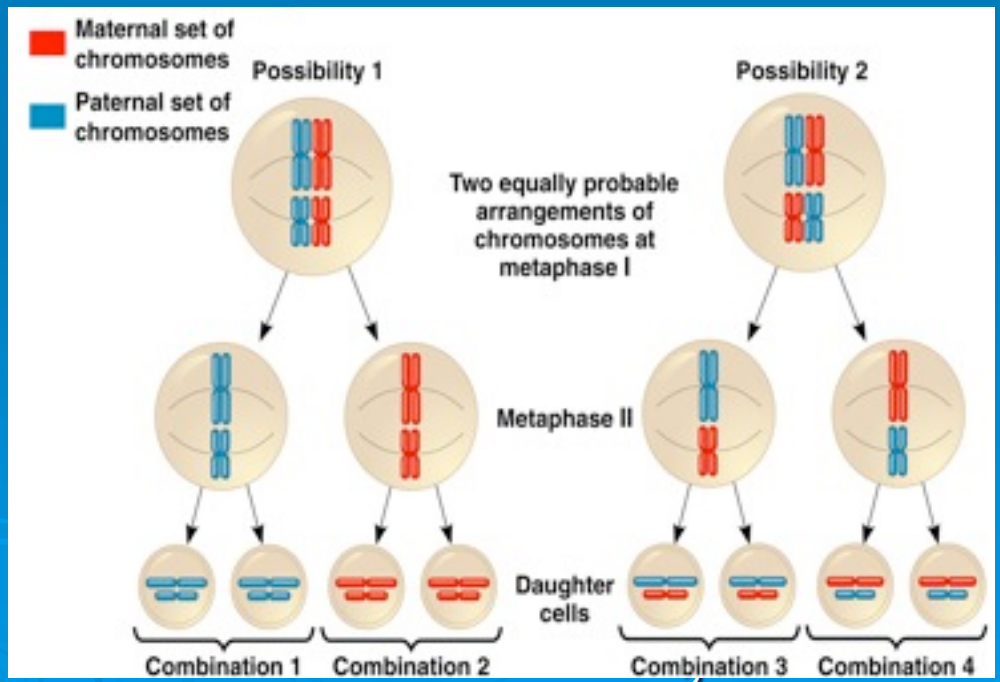
Topic 4 & 10: Genetics

How does Meiosis create genetic variation?

- Crossing over

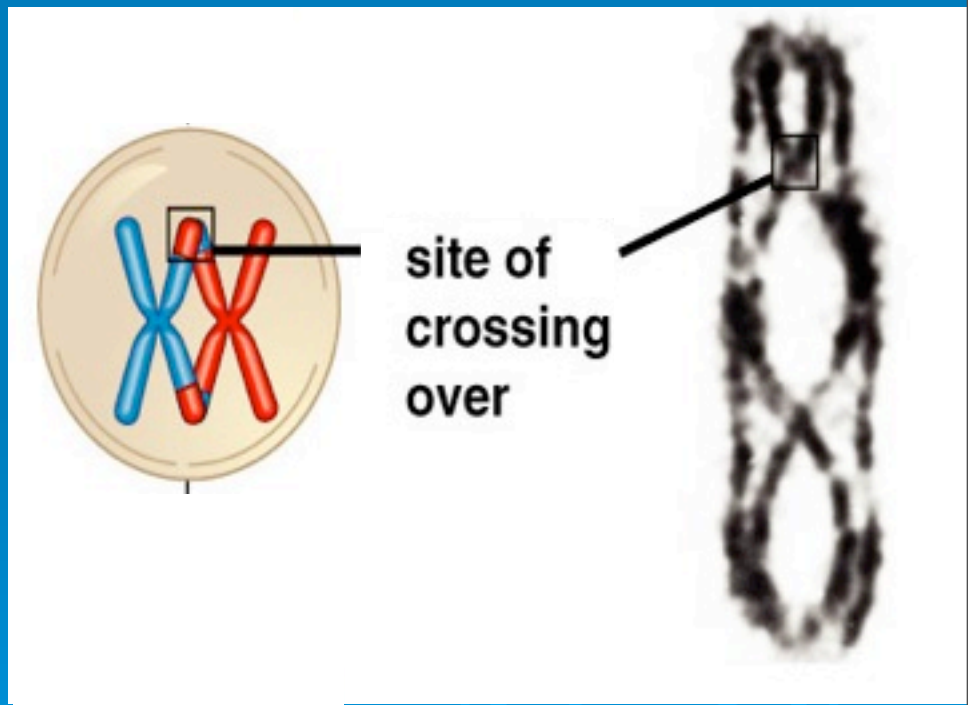


- Independent assortment



Crossing Over

- an exchange of parts of non-sister chromatids
- occurs at chiasmata of joined tetrads

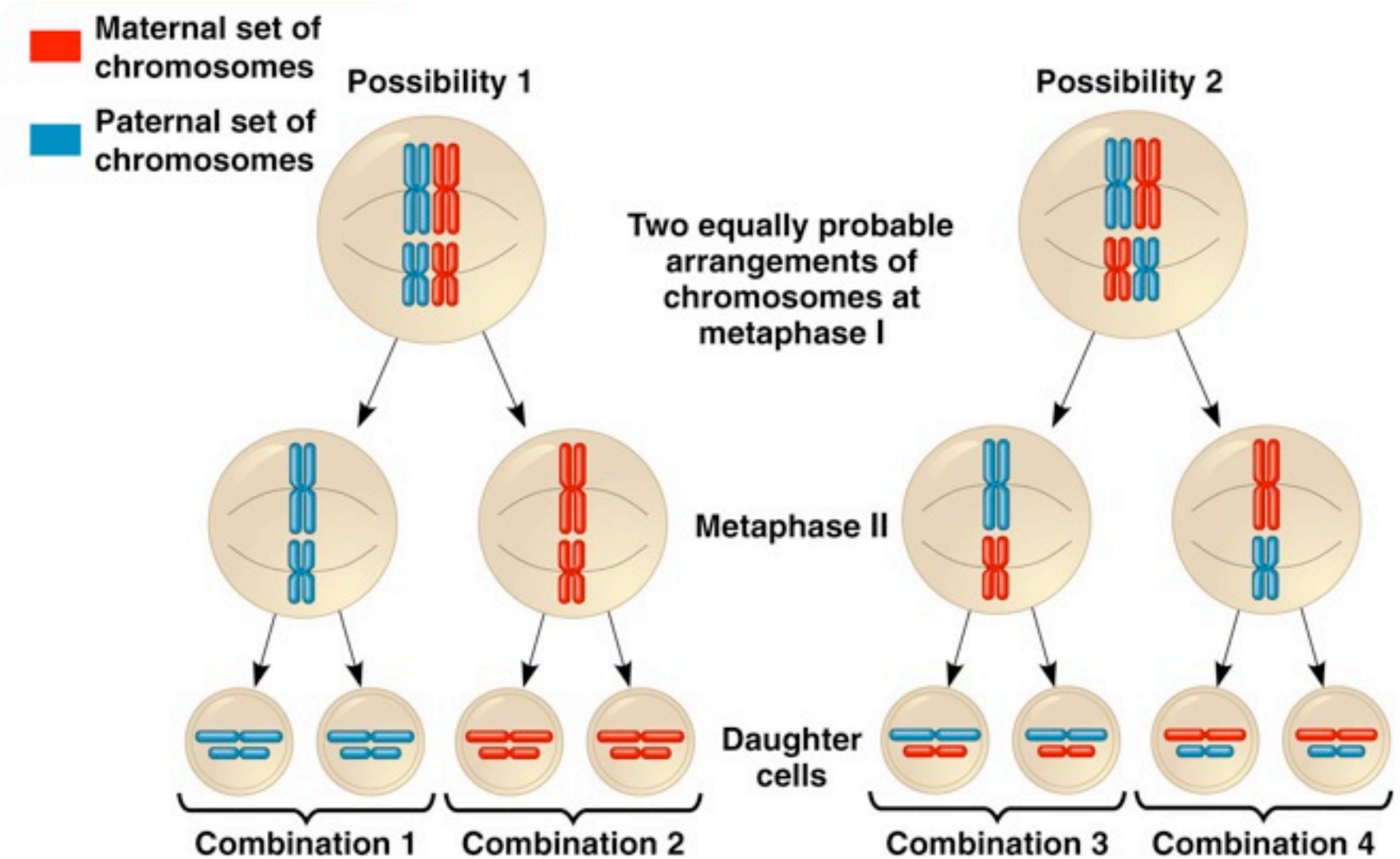


- Happens during Prophase I of meiosis
- One of the main sources of genetic variation

Independent Assortment

- another main source of genetic variation
- it is the random alignment of maternal/ paternal chromosomes at the metaphase plate

Independent Assortment

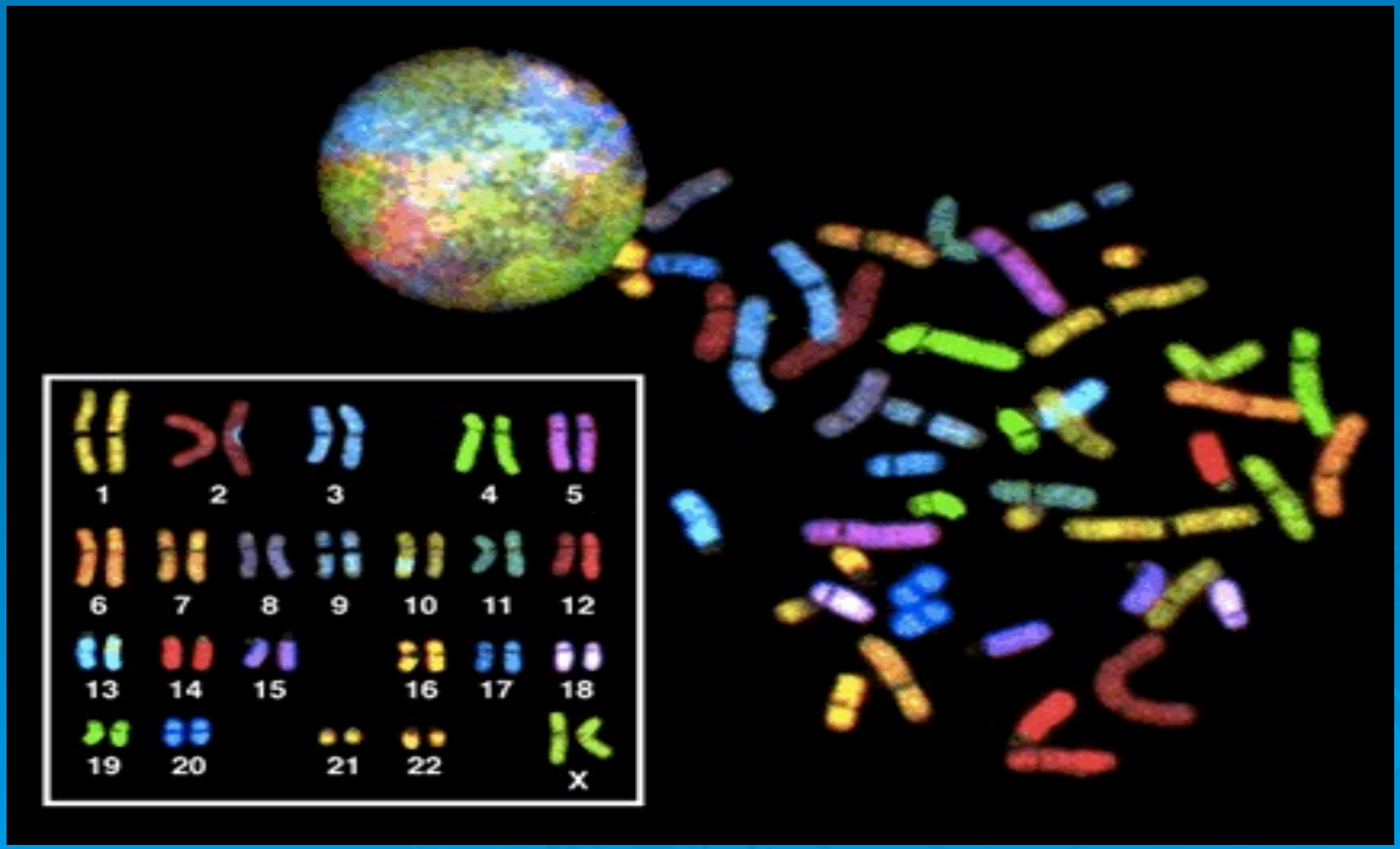


Karyotypes and Nondisjunction

What is a Karyotype?

- **Karyotyping** is performed using cells collected by chorionic villus sampling or amniocentesis, for pre-natal diagnosis of chromosome abnormalities.
- A picture of chromosomes arranged in homologous pairs by
  - size (biggest to smallest)
  - shape
  - centromere position
- Many karyotypes are used to identify abnormal amount of chromosomes or to find out the sex of a person

Spectral Karyotype of a Human



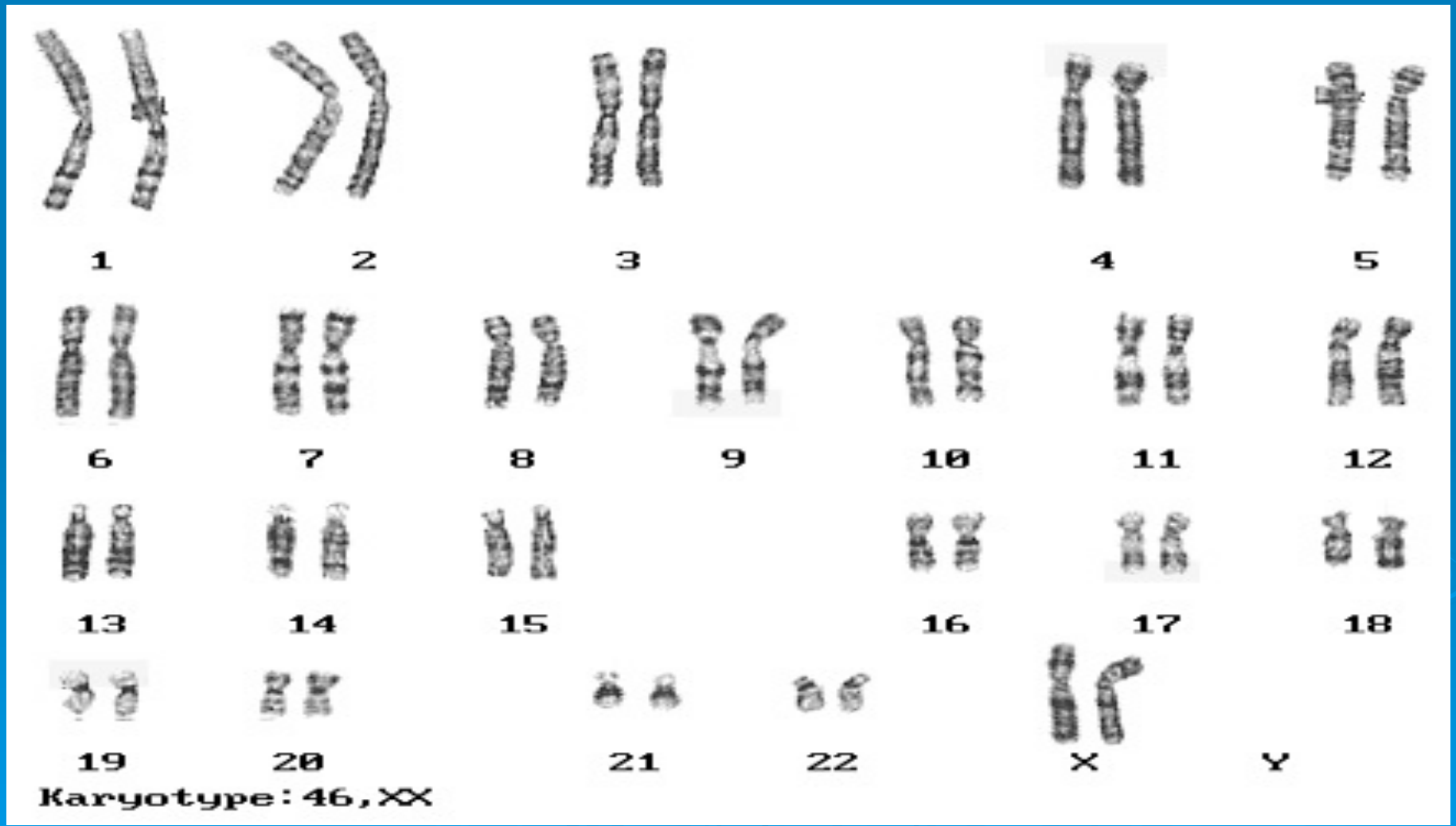
Why do a Karyotype?

- Help to determine genetic abnormalities (also sex)
- Leads to many ethical concerns:
  - Balancing the risks of side-effects (for example, miscarriage) agains the possibility of identifying and aborting a fetus with an abnormality.
  - Who should make the decision about whether to perform karyotyping and allow a subsequent abortion—parents or health-care professionals or both groups?
  - Whether or not national governments should interfere with personal freedoms, and whether or not they should be able to ban procedures within the country and possibly also ban citizens travelling to foreign countries where the procedures are permitted.

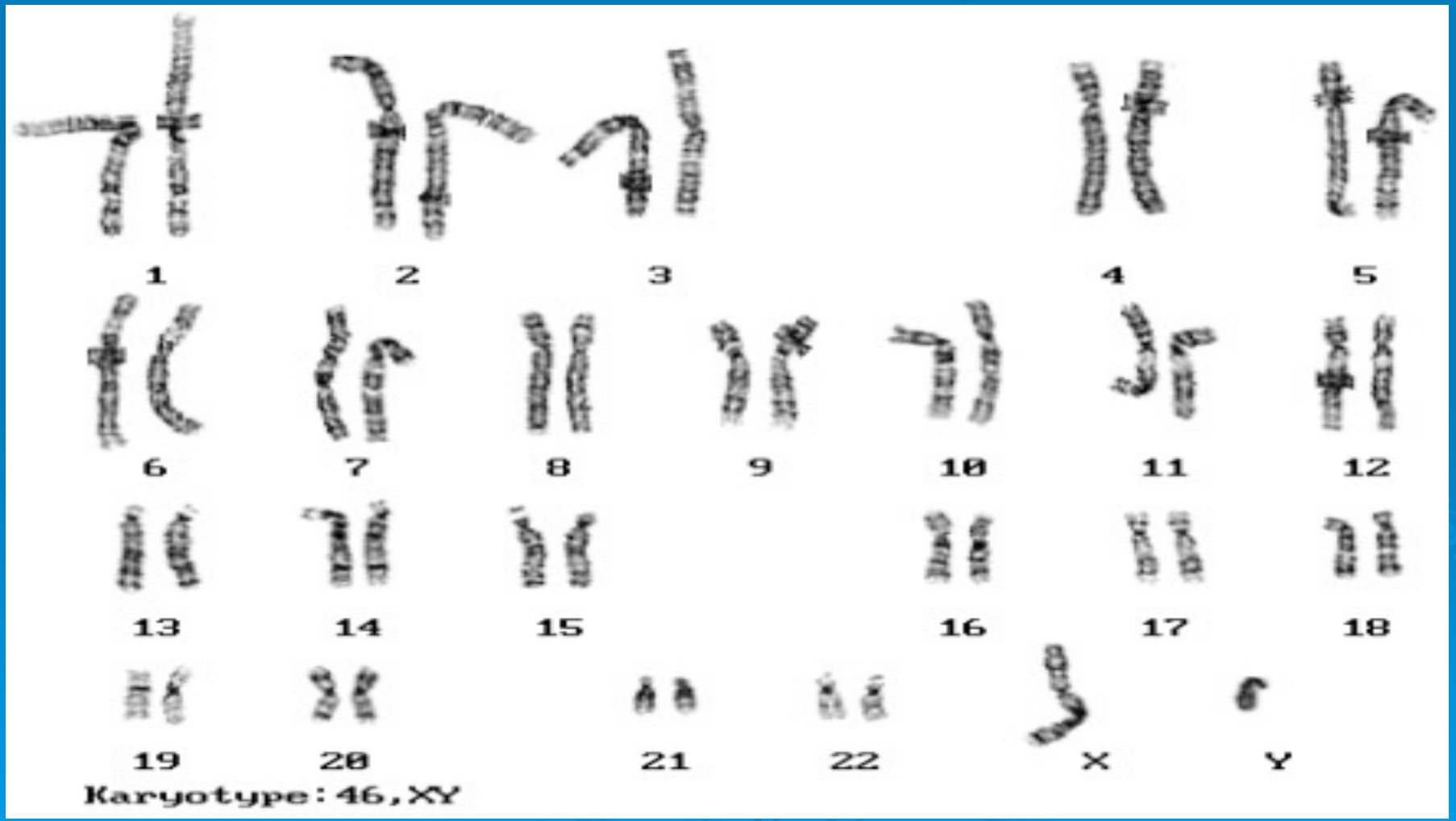
Four Corners

- Would you have a KARYOTYPE?
- What if you had a “high risk” pregnancy?

Female Karyotype
(notice there are two X chromosomes)

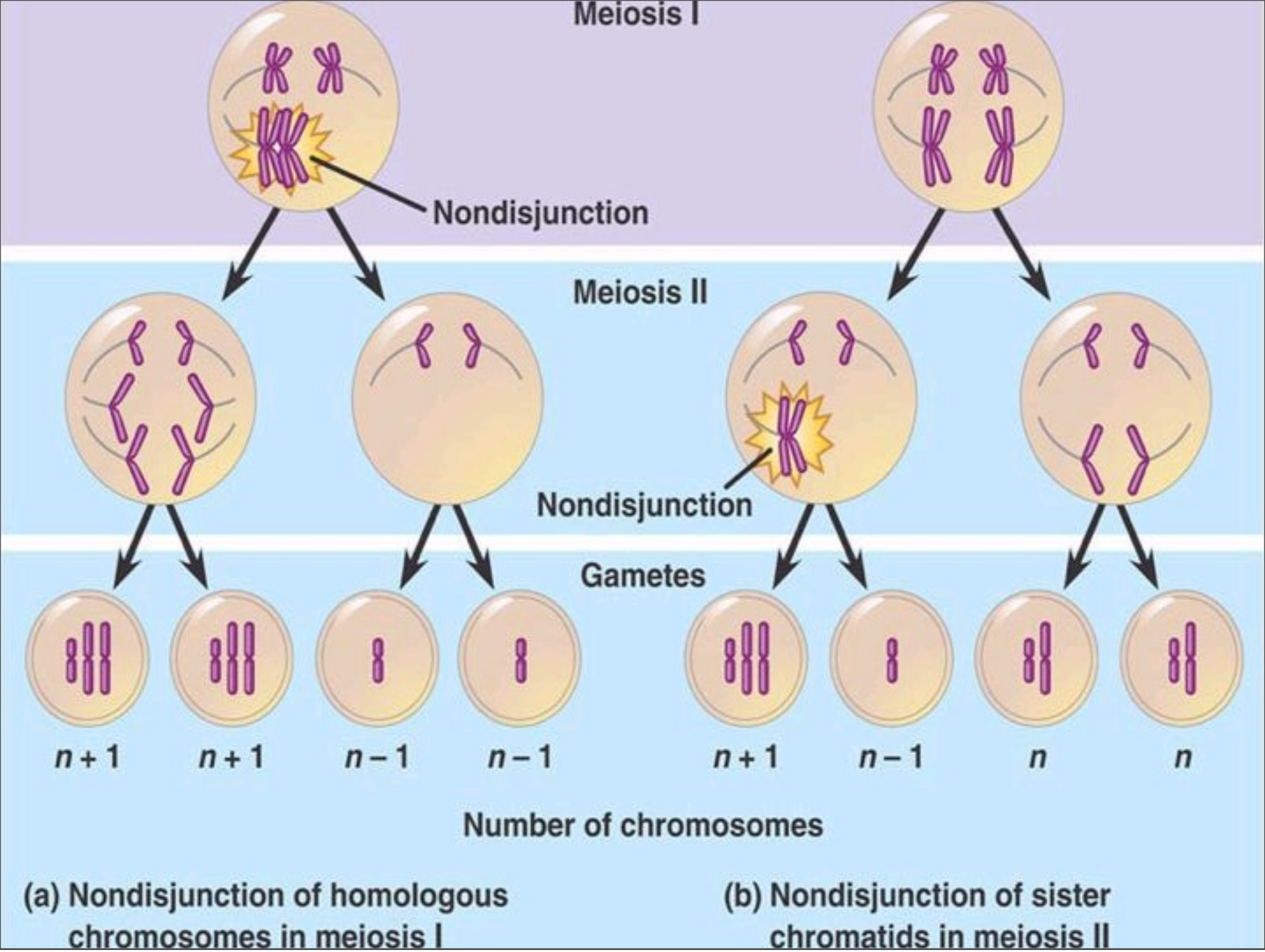


Normal Male Karyotype (Notice that there is an X and Y chromosome)

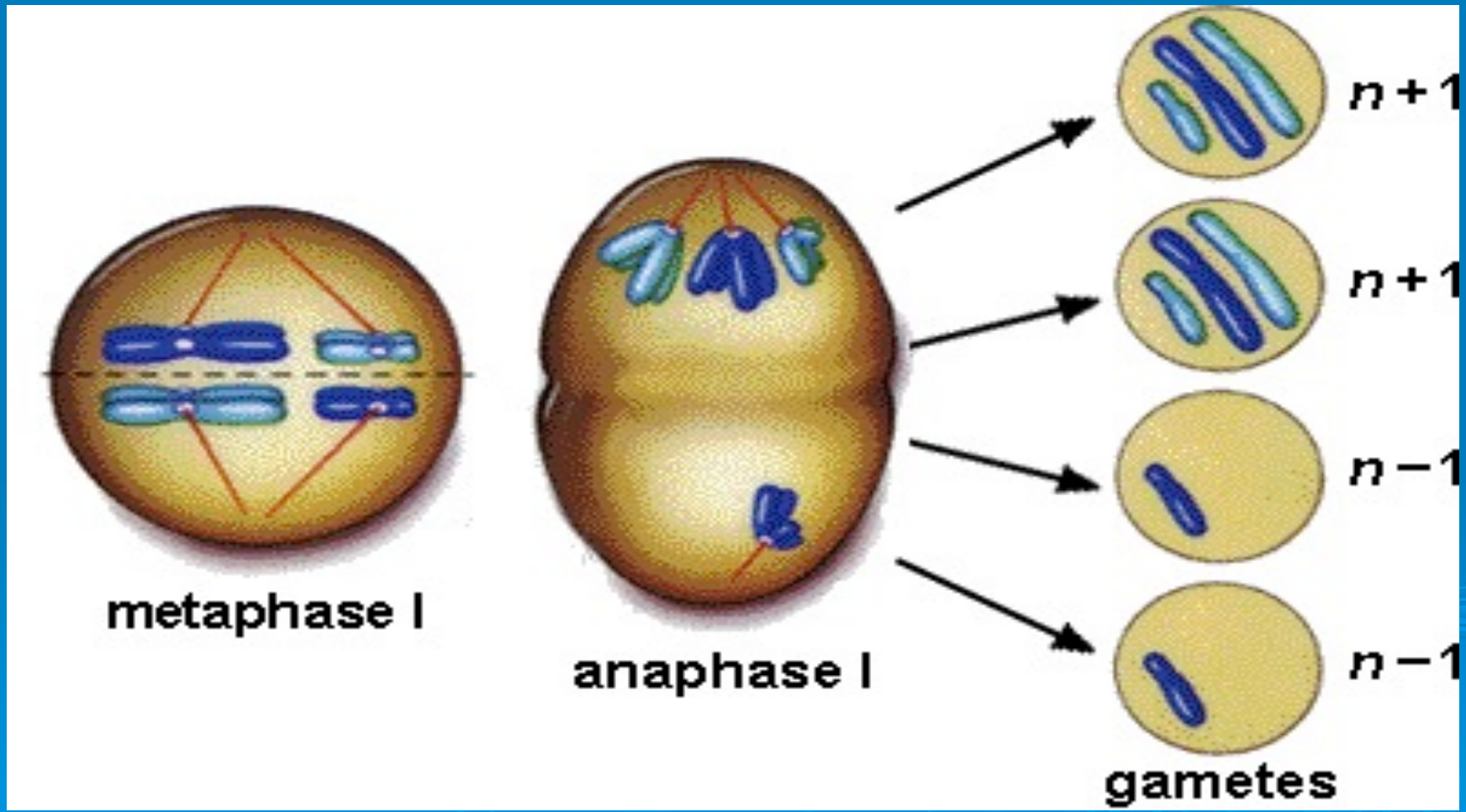


How Nondisjunction Occurs

- Nondisjunction occurs when either:
  - Homologues fail to separate during anaphase I of meiosis
  - Sister chromatids fail to separate during anaphase II of meiosis
- Result : one gamete has 2 copies of one chromosome & other has no copy of that chromosome (the other chromosomes are distributed normally.)
- During fertilization, the result is aneuploidy (abnormal chromosome number)


Meiosis I
Nondisjunction
Meiosis II
Nondisjunction
Gametes
n + 1
n + 1
n – 1
n – 1
n + 1
n – 1
n
n
Number of chromosomes
(a) Nondisjunction of homologous chromosomes in meiosis I
(b) Nondisjunction of sister chromatids in meiosis II

Homologues Fail to Separate During Anaphase I of Meiosis



Types of Nondisjunction

- **Trisomic** cell has one extra chromosome (2n +1) = example: trisomy 21. (Polyploidy refers to the condition of having three homologous chromosomes rather then two)
- **Monosomic** cell has one missing chromosome (2n - 1) = usually lethal except for one known in humans: Turner's syndrome (monosomy)

Can Nondisjunction Infants Survive?

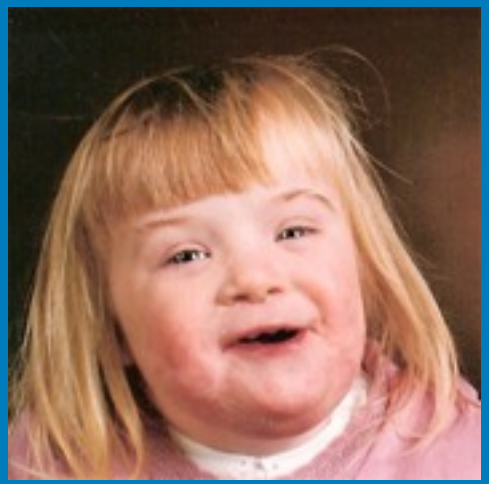
- Frequency of nondisjunction is quite high in humans, but results are usually so devastating to the growing zygote that miscarriage occurs very early in the pregnancy
- If the individual survives, he or she usually has a set of symptoms - a syndrome - caused by the abnormal dose of each gene product from that chromosome.

Chromosomal Abnormalities

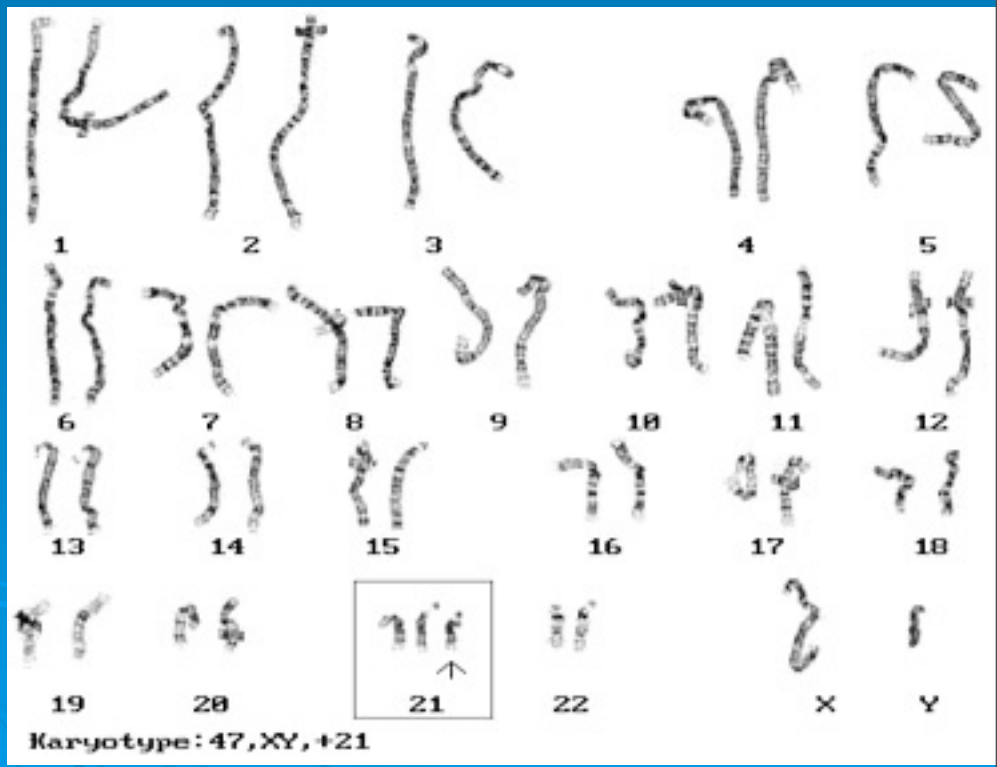
Name	Chromosome Annotation
Down Syndrome	Trisomy 21
Edward's Syndrome	Trisomy 18
Patau's Syndrome	Trisomy 13
Kleinfelter's Syndrome	47 chromosomes; XXY
XXY	47 chromosomes
XXX	47 chromosomes
XO	45 chromosomes

What's the Problem?

Down's Syndrome (autosomal)

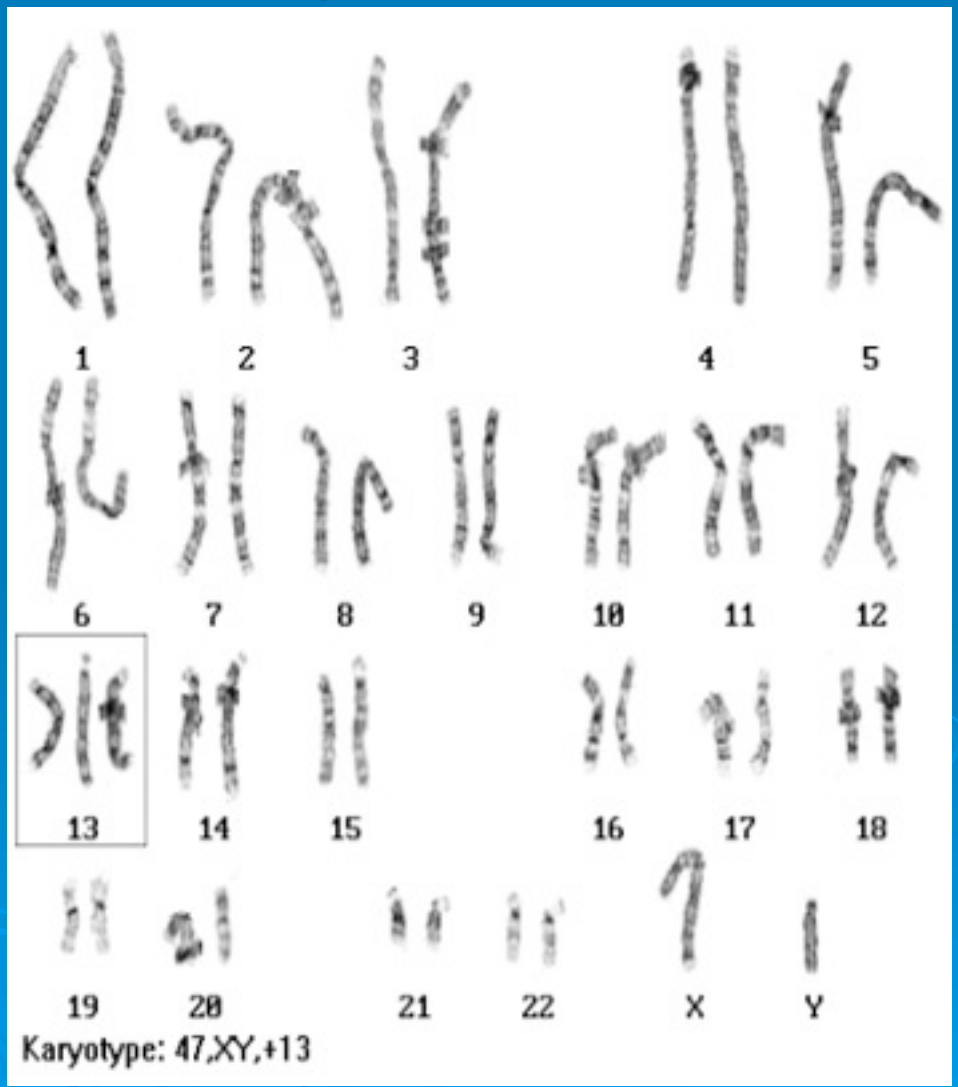


- Children with Down syndrome have an extra chromosome 21 (Trisomy 21
- Characteristics:
  - Round face
  - Mental impairment
  - Slanted eyes
  - Flattened Forehead
  - Underdeveloped reproductive organ

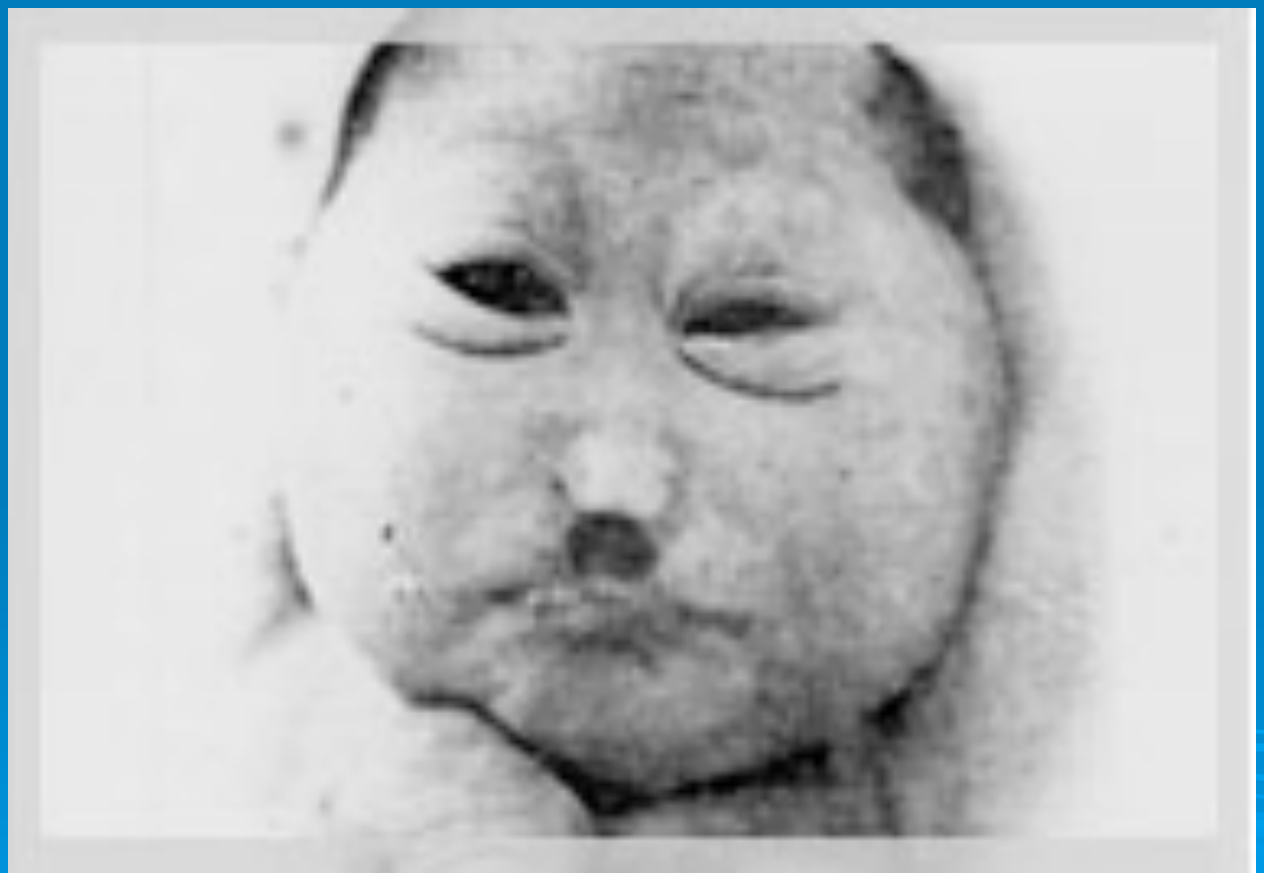


Patau's Syndrome (autosomal)

- 47 chromosomes trisomy 13
- Characteristics:
- serious eye, brain, circulatory defects as well as cleft palate
- 1:5000 live births.
- rarely live more than a few months

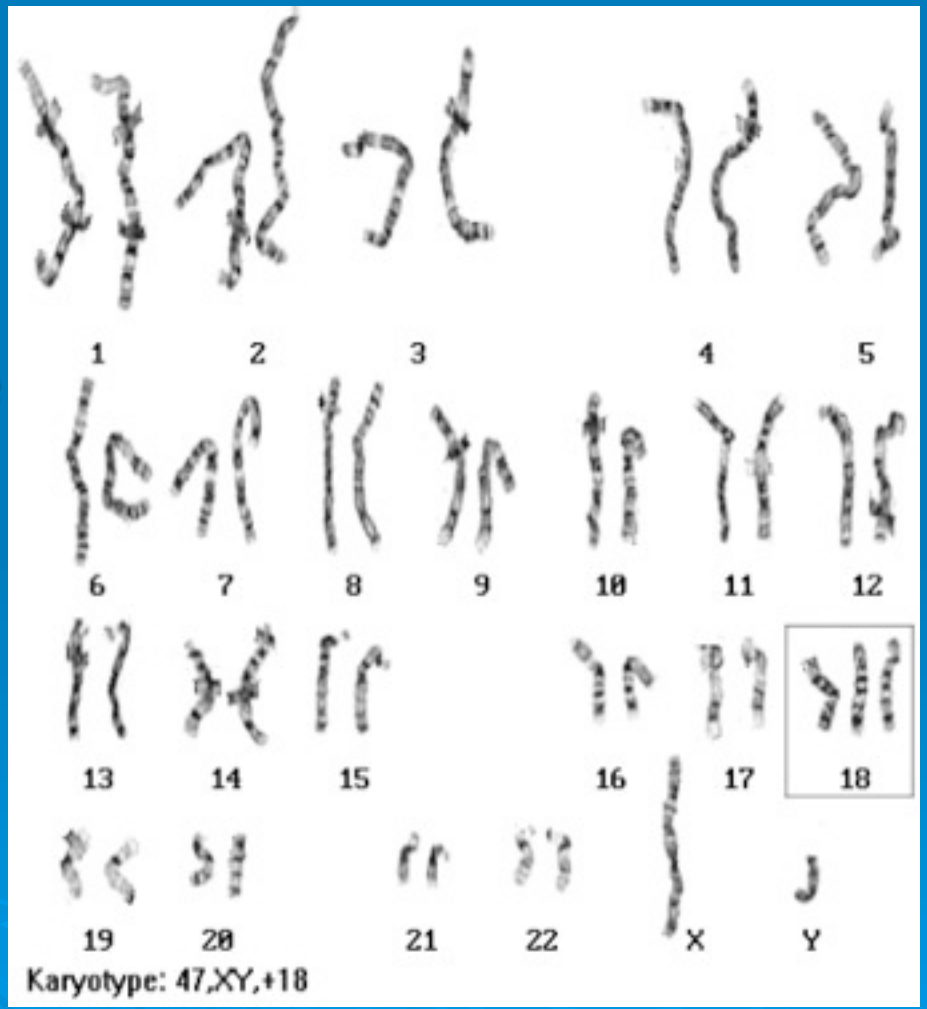


Patau's Syndrome



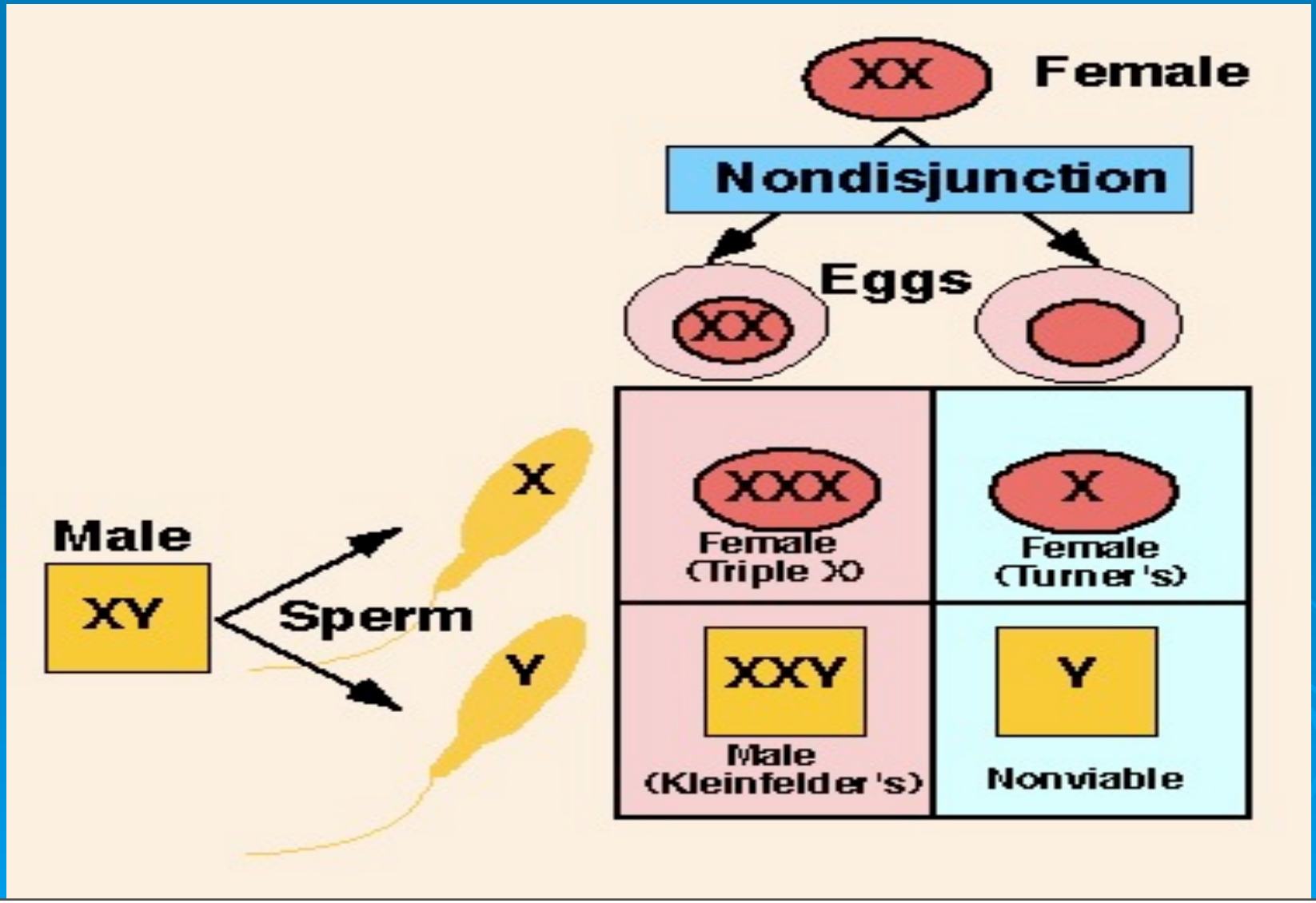
Edward's Syndrome (autosomal)

- 47 chromosomes, trisomy 18
- almost every organ system affected
- Characteristics: severe mental impairment, multiple deformities (skull, heart & kidney defects, club-feet)
- 1:10,000 live births
- Generally do not live more than a few months



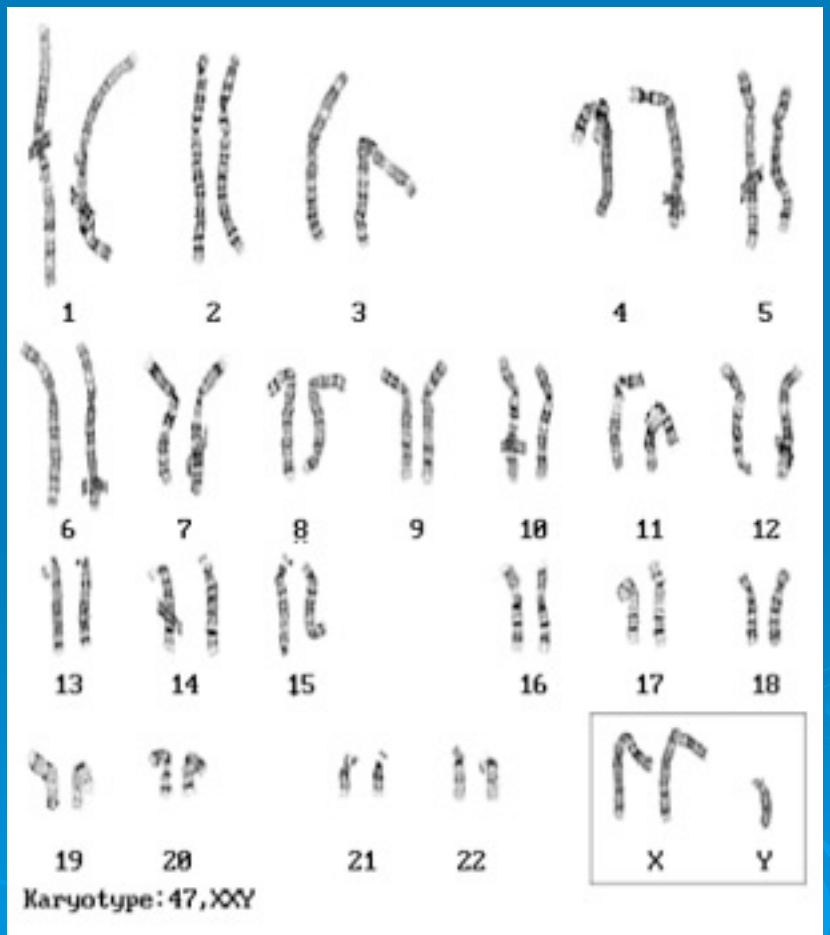
Edward's Syndrome

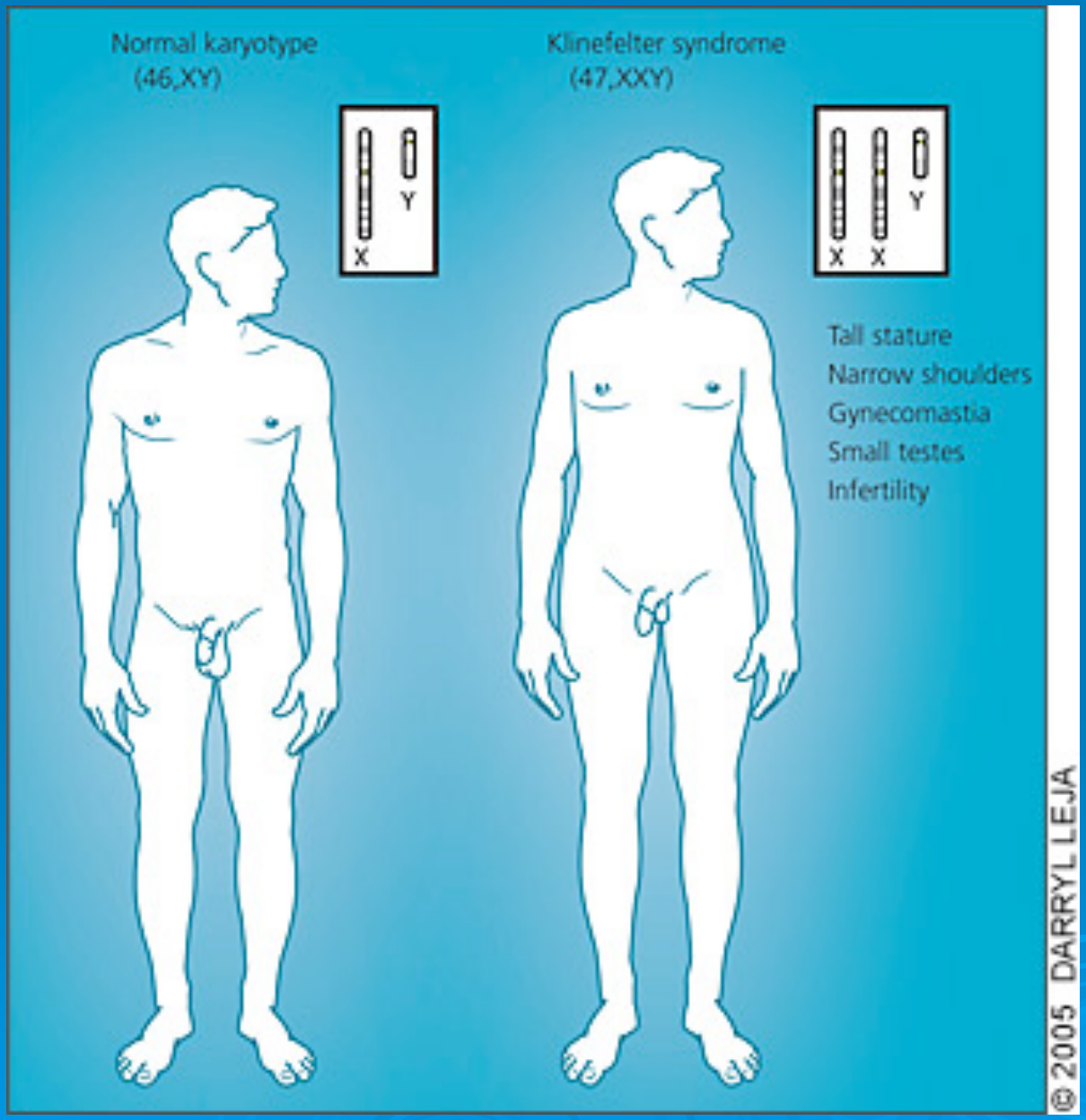
Nondisjunction and Associated XY Disorders



Klinefelter's Syndrome (sex chromosome)

- 47 chromosomes XXY
- unusually small testes, sterile
- Breast enlargement and other feminine body characteristics
- normal intelligence




Normal karyotype
(46,XY)
X
Y
Klinefelter syndrome
(47,XXY)
X
X
Y
Tall stature
Narrow shoulders
Gynecomastia
Small testes
Infertility
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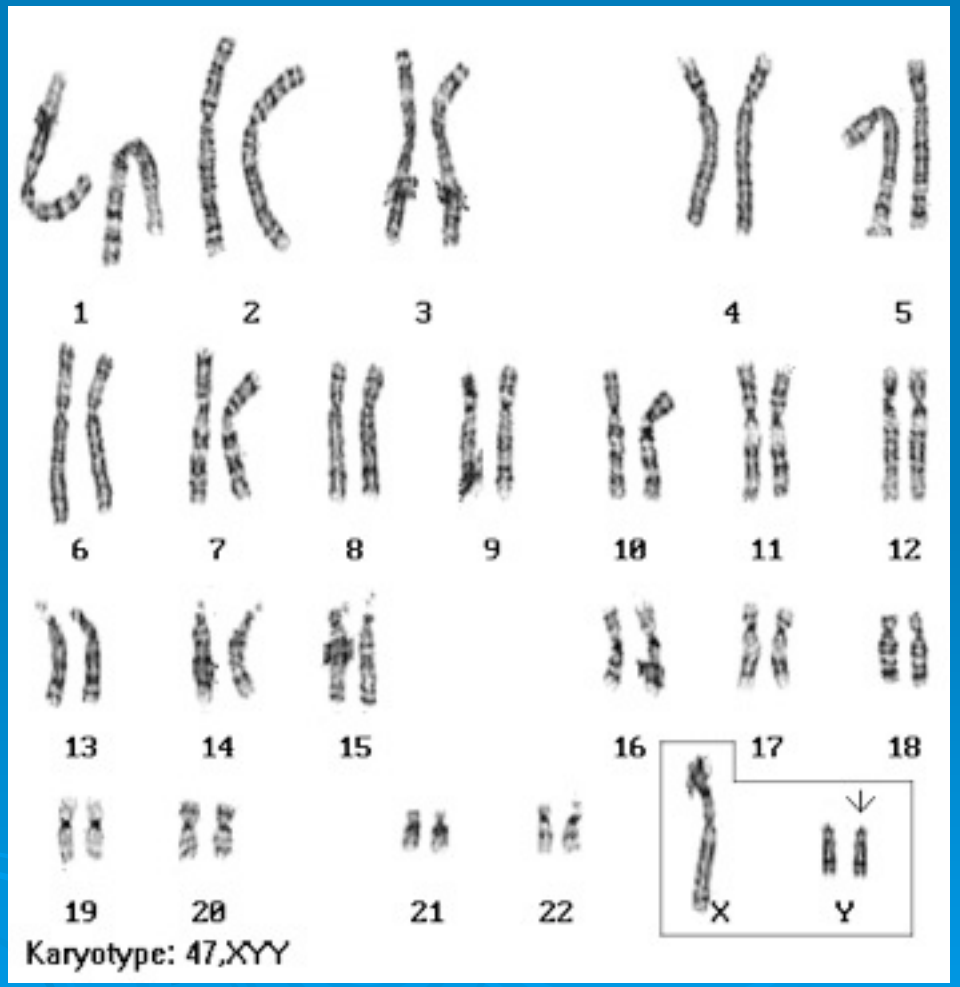
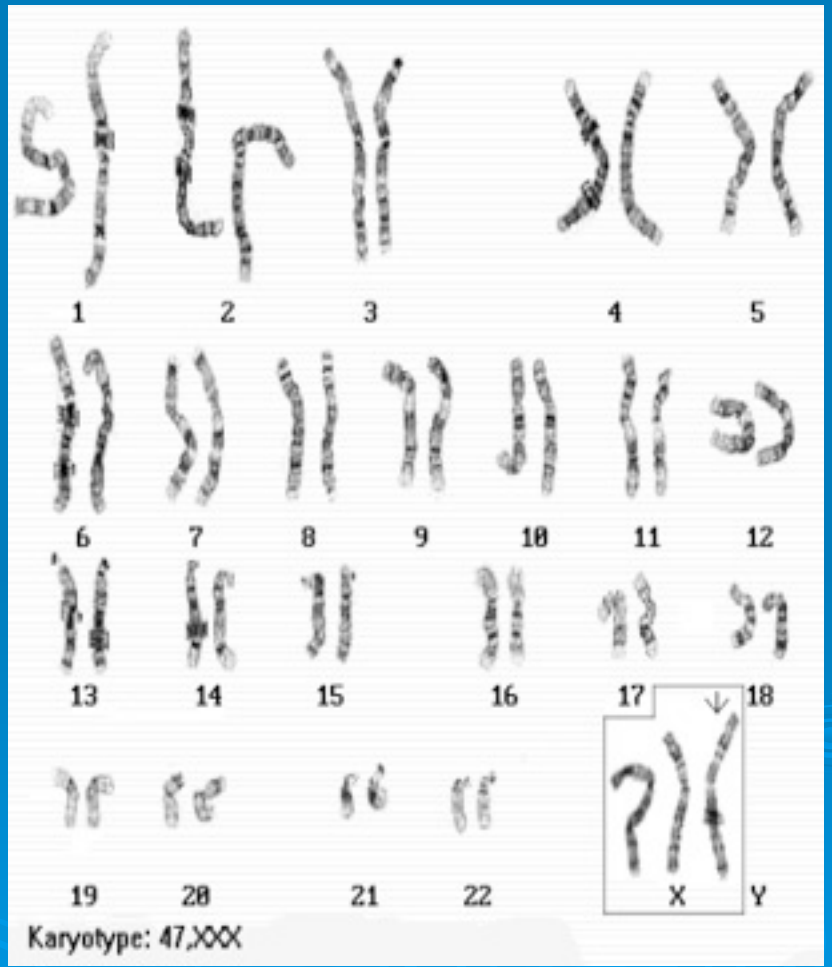
“Supermale” (sex chromosome)

- 47 chromosomes (XYY)
- Individuals are somewhat taller than average
- often below normal intelligence
- it was thought that these men were likely to be criminally aggressive, but this hypothesis has been disproven over time.



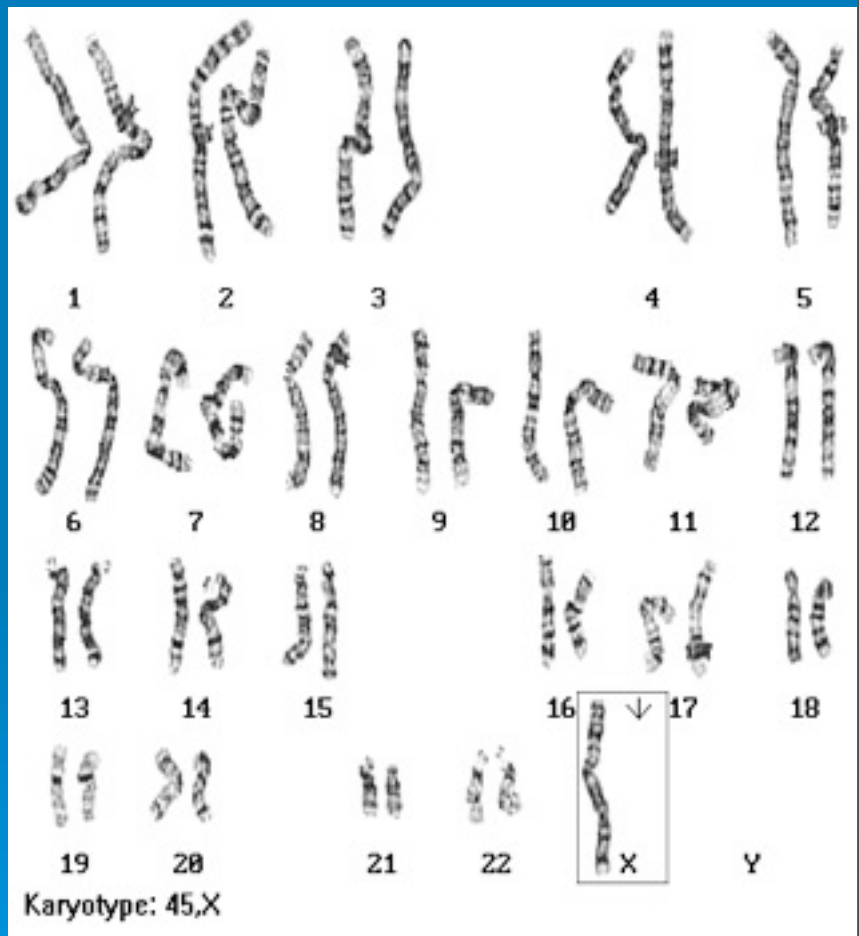
Superfemales (sex chromosomes)

- 47 chromosomes (XXX)
- healthy and usually fertile
- cannot be distinguished from normal female except by karyotype

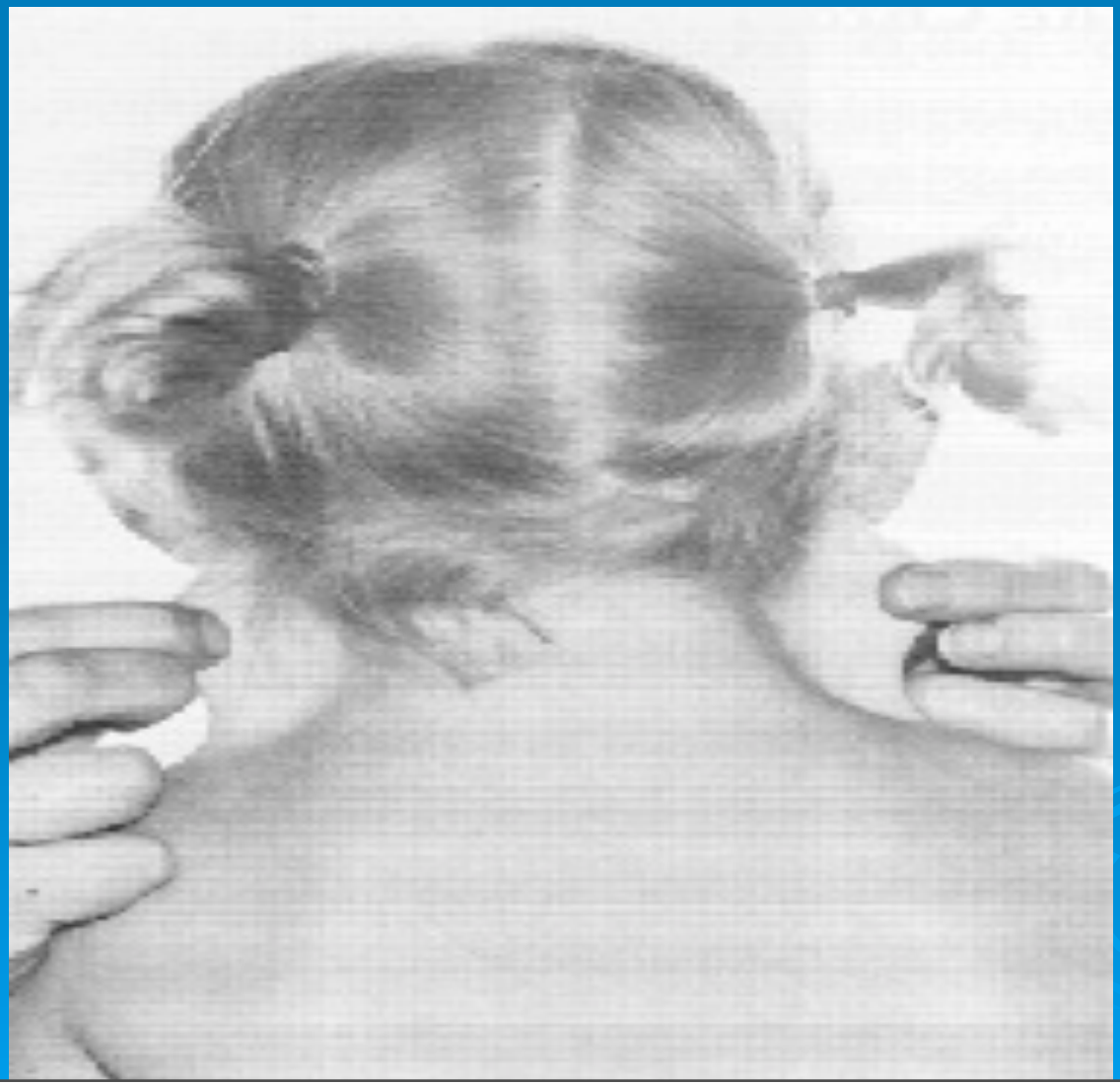


Turner's Syndrome

- 45 chromosomes (one X)
- 1:5000 live births
- the only viable monosomy in humans - women with Turner's have only 45 chromosomes
- XO individuals are genetically female, however, they do not mature sexually during puberty and are sterile
- Short stature and normal intelligence. (98% of these fetuses die before birth)



Turner's Syndrome



- http://www.biology.iupui.edu/biocourses/N100/2k2humancsomaldisorders.html