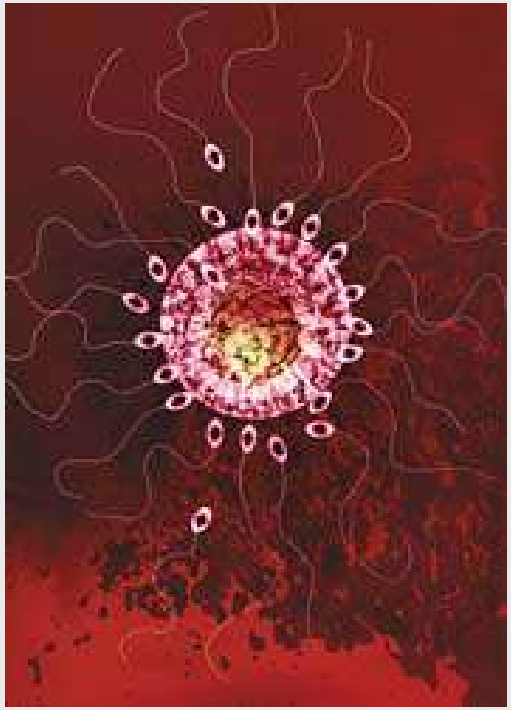
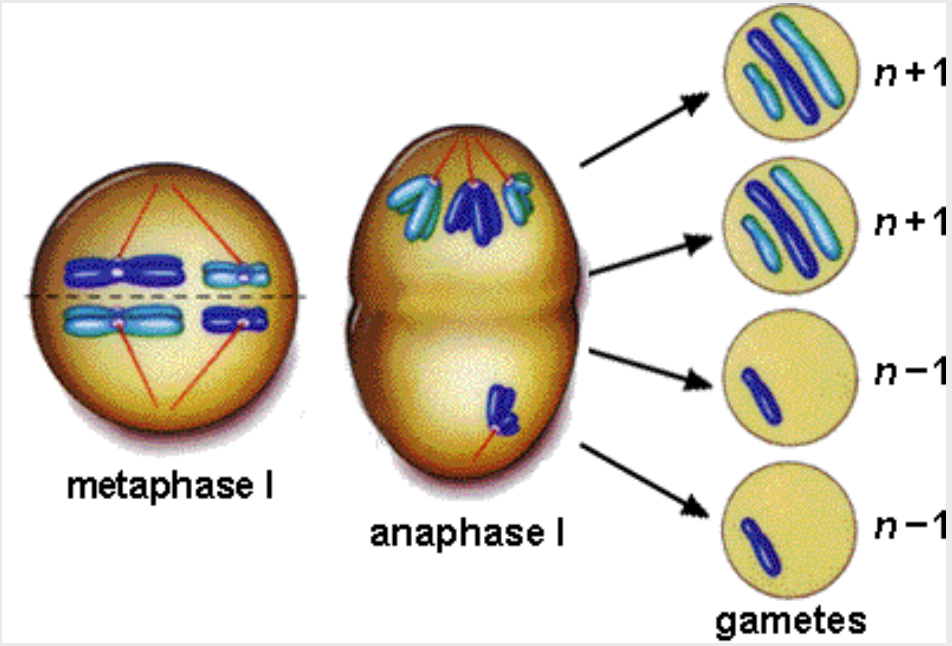
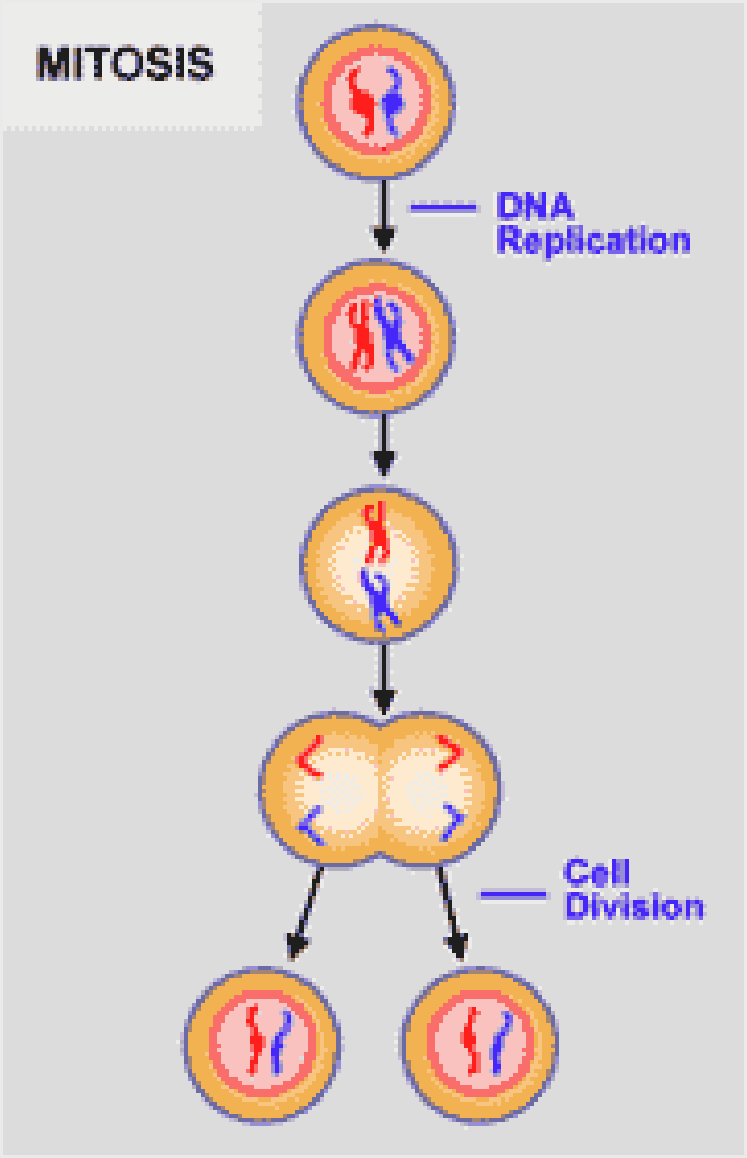
NOTES - MEIOSIS:
Genetic Variation / Mistakes in Meiosis

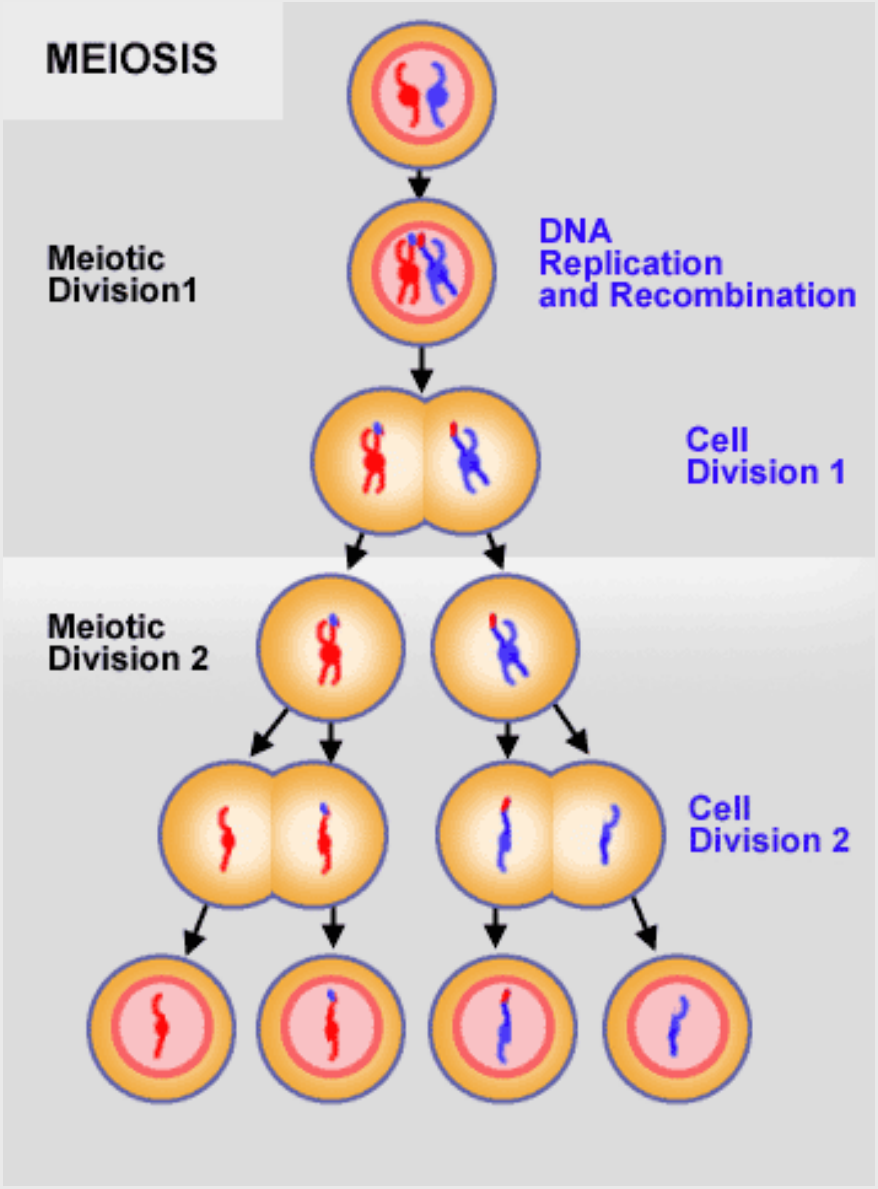


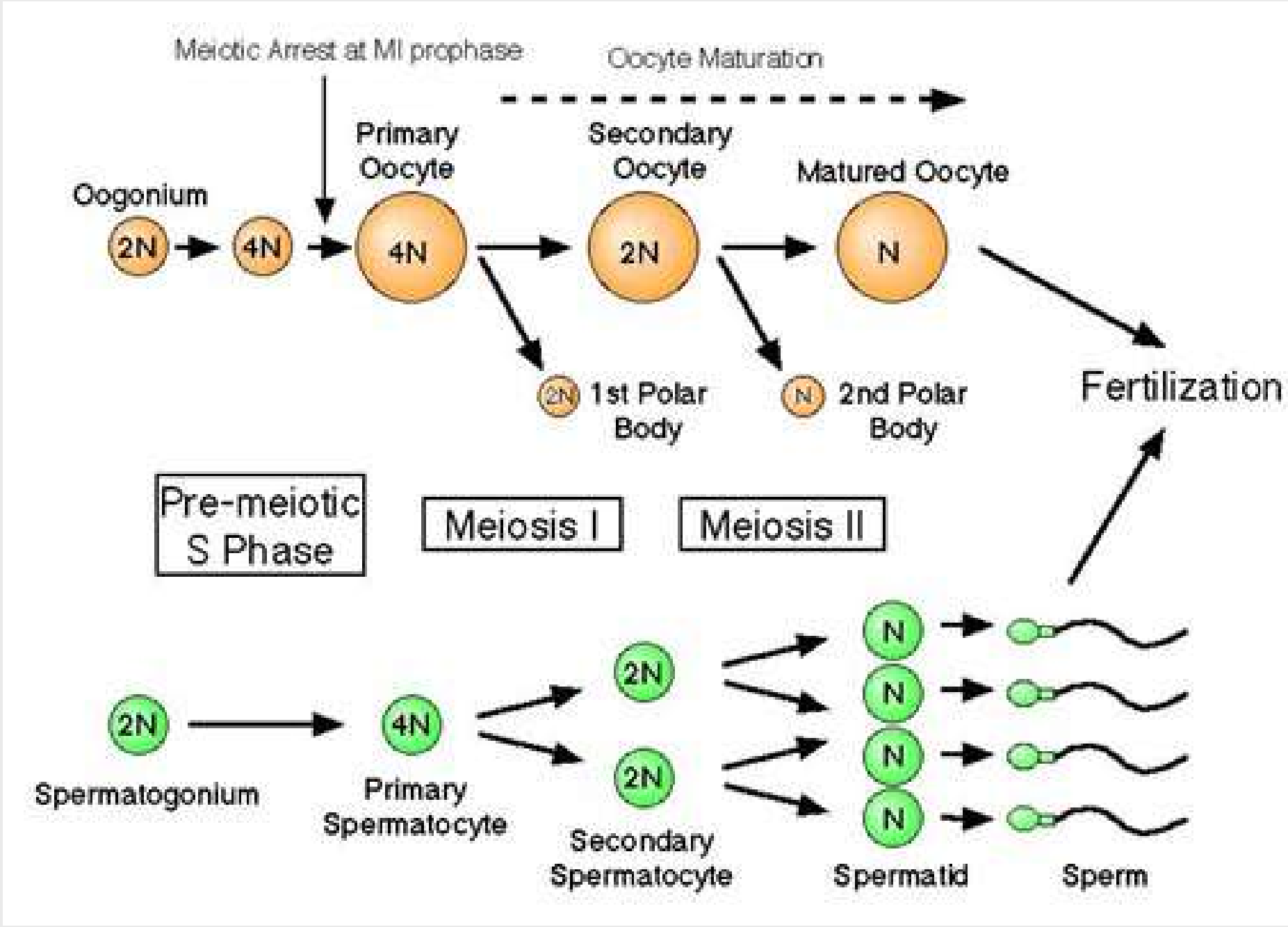


RECALL: Mitosis and Meiosis differ in several key ways:

MITOSIS:	**MEIOSIS:**
1 round of cell division	2 rounds of cell division
Produces **2 DIPLOID** daughter cells	Produces **4 HAPLOID** daughter cells
Daughter cells are **identical** to parent cell	Daughter cells are **different** from parent cell AND from each other
Produces **body cells**	Produces **GAMETES** (sperm and egg cells)

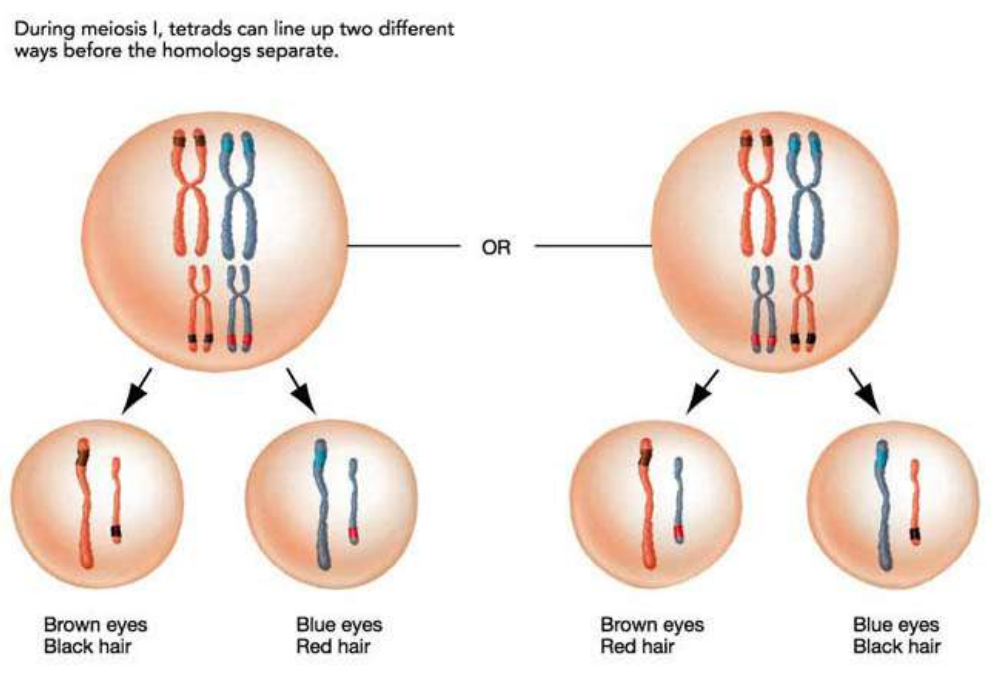

MITOSIS
DNA Replication
Cell Division


MEIOSIS
Meiotic Division1
DNA Replication and Recombination
Cell Division 1
Meiotic Division 2
Cell Division 2


Meiotic Arrest at MI prophase
Oocyte Maturation
Primary Oocyte
Secondary Oocyte
Matured Oocyte
Oogonium
2N
4N
4N
2N
N
2N 1st Polar Body
N 2nd Polar Body
Fertilization
Pre-meiotic S Phase
Meiosis I
Meiosis II
2N
4N
2N
2N
N
N
N
N
Spermatogonium
Primary Spermatocyte
Secondary Spermatocyte
Spermatid
Sperm

How does MEIOSIS lead to GENETIC VARIATION?

1) RANDOM ASSORTMENT: chromosome "shuffling"; how the chromosomes line up in metaphase I

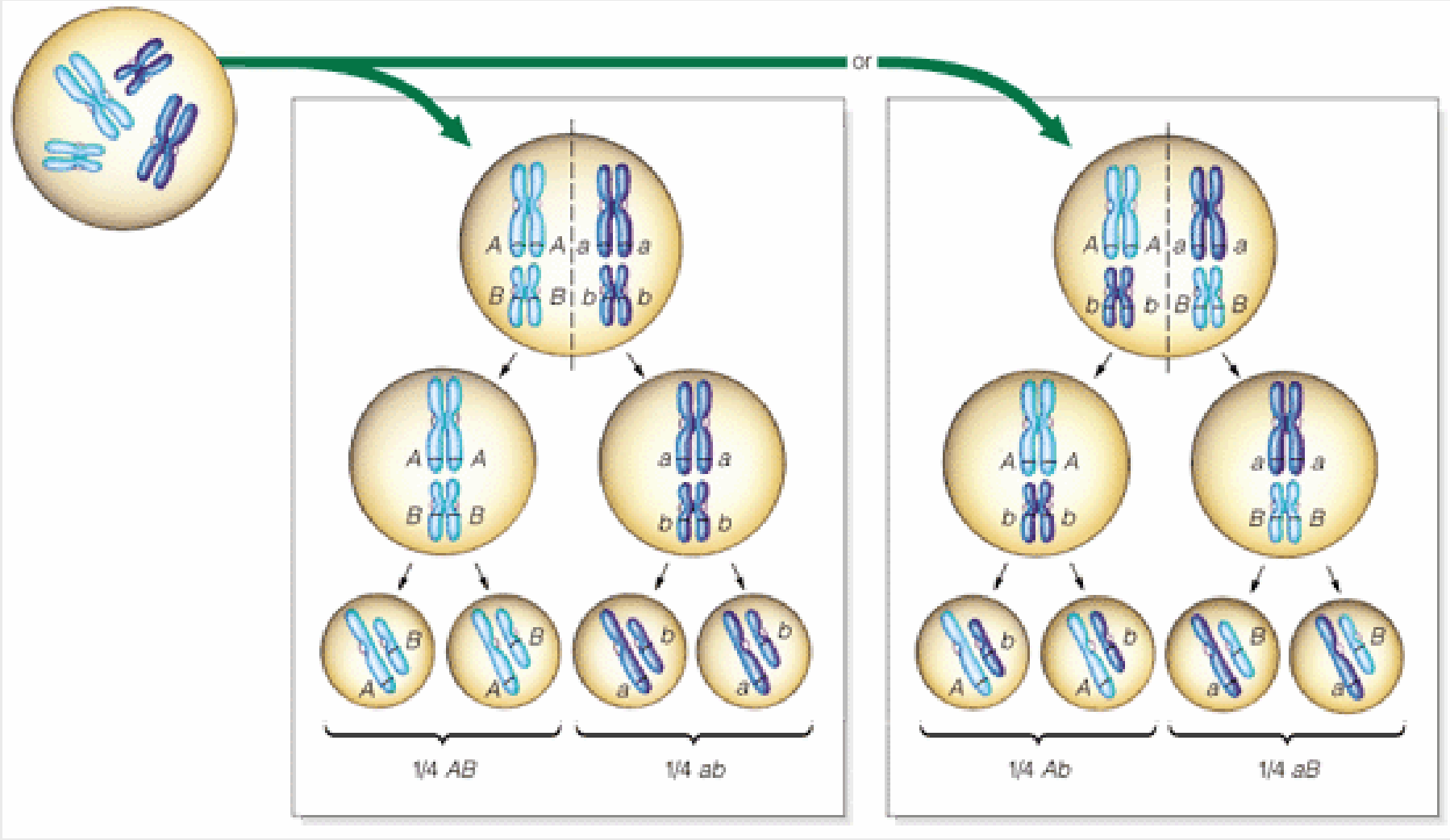


CHROMOSOME SHUFFLING:

Example: in pea plants, there are 7 pairs of chromosomes; each of the 7 pairs can line up in 2 different ways…this means that there are 128 different sperm or egg cells possible!

(2^7=128)

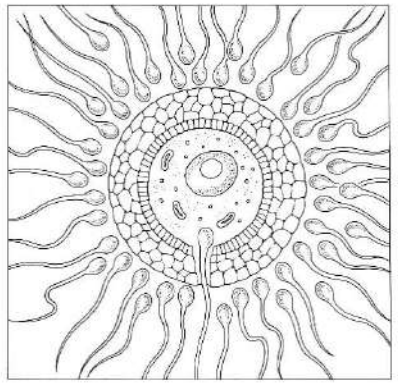
the lining up of homologous pairs of chromosomes during metaphase I is a* ***RANDOM PROCESS*!*


or
A A a a
B B b b
A A
B B
a a
b b
A B
a b
1/4 AB
1/4 ab
A A a a
b b B B
A A
b b
a a
B B
A b
a B
1/4 Ab
1/4 aB

CHROMOSOME SHUFFLING:

And…since there are 128 different possible sperm cells, and 128 different possible egg cells, the # of possible combinations is:

128 x 128 = 16,384!



CHROMOSOME SHUFFLING:

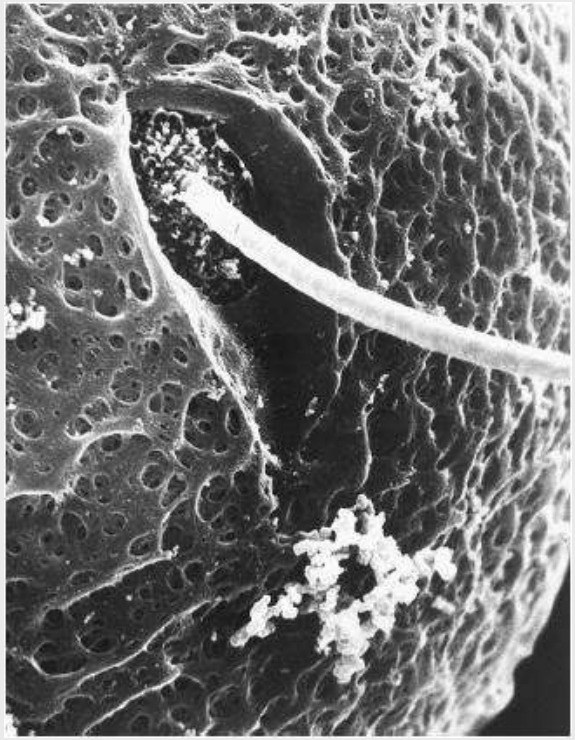
**So how many combinations are possible in humans???

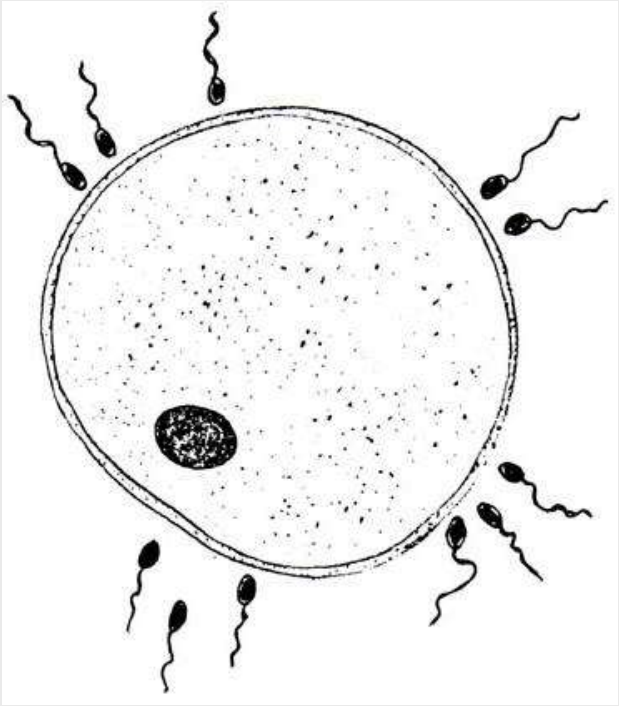
Answer:

2^{23}=**8,388,608 different** egg or sperm cells…and…

8,388,608 x 8,388,608 =

Over **70 trillion** **different zygotes** possible!



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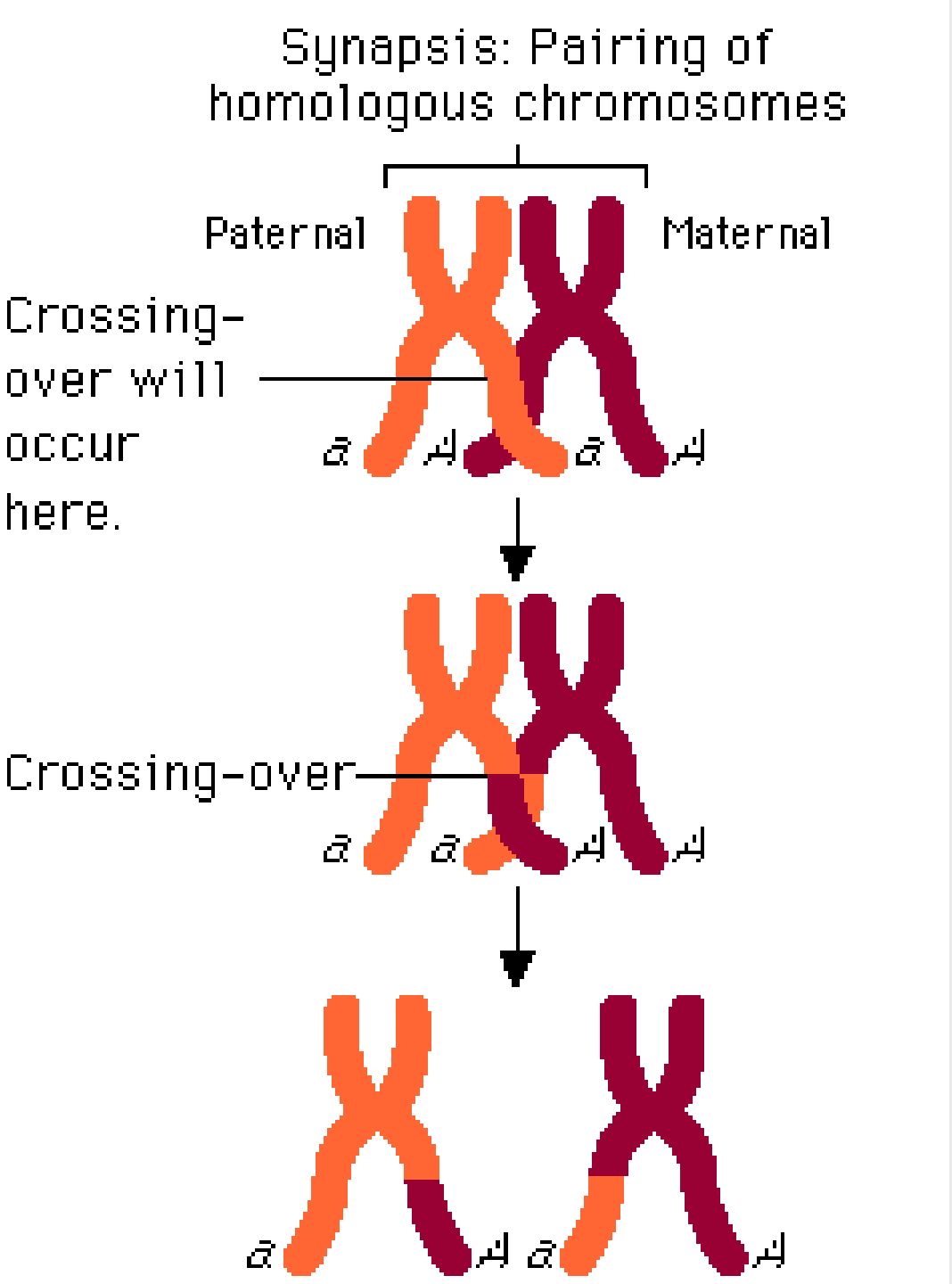
How does MEIOSIS lead to GENETIC VARIATION?

2) CROSSING OVER:

-occurs in **prophase I** when chromosomes line up with their homologous partner

-chromosome segments are **exchanged** between homologous chromosomes

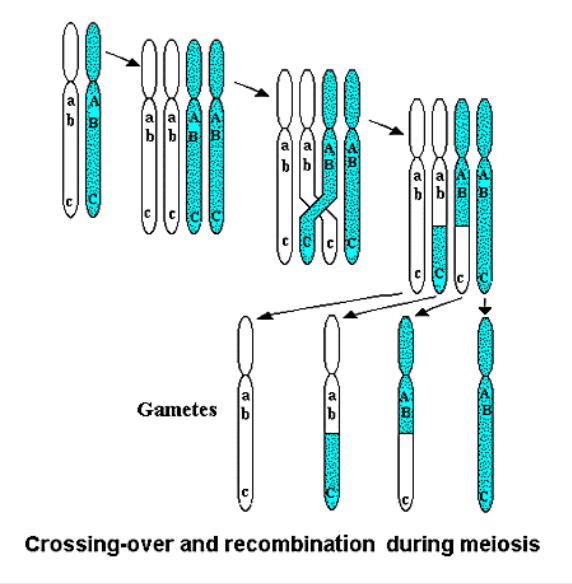
-produces brand new combinations of genes on chromosomes


Synapsis: Pairing of homologous chromosomes
Paternal
Maternal
Crossing-over will occur here.
a
a
Crossing-over
a
a
a
a

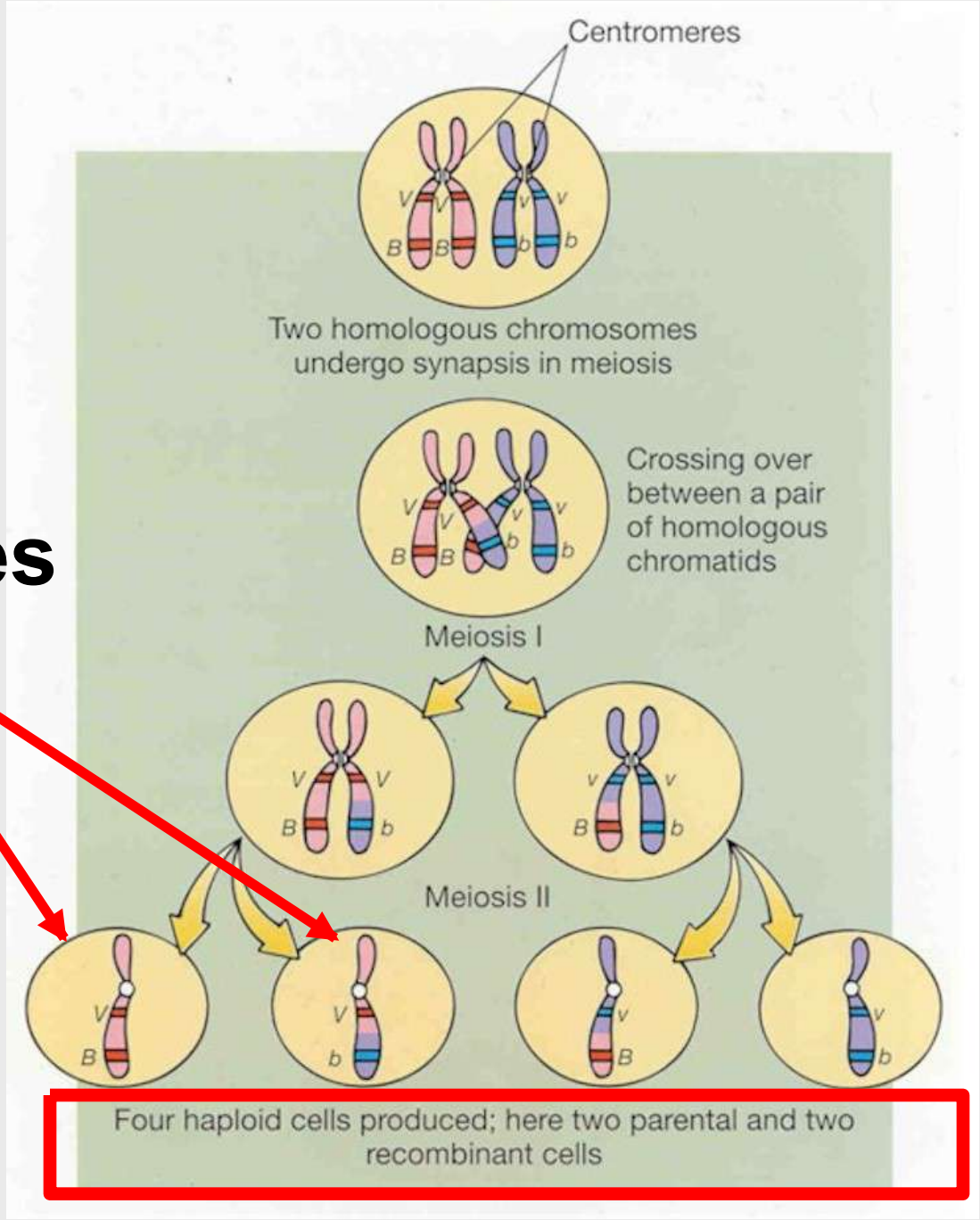
CROSSING OVER:

-typically, there are 2 or 3 crossovers per chromosome pair…

-this means an almost endless # of different possible chromosomes!



Gametes


Centromeres
Two homologous chromosomes undergo synapsis in meiosis
Crossing over between a pair of homologous chromatids
Meiosis I
Meiosis II
Four haploid cells produced; here two parental and two recombinant cells

GENETIC RECOMBINATION:

- this reassortment of chromosomes and genetic information as a result of:

-independent segregation ("shuffling")

-crossing over

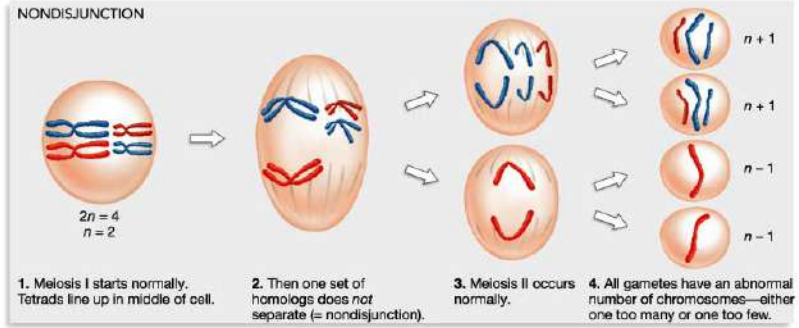
- a major source of **variation** among organisms;
- the "raw material" that forms the basis for evolution (natural selection!)

NOVA video

http://www.pbs.org/wgbh/nova/baby/divi_flash.html

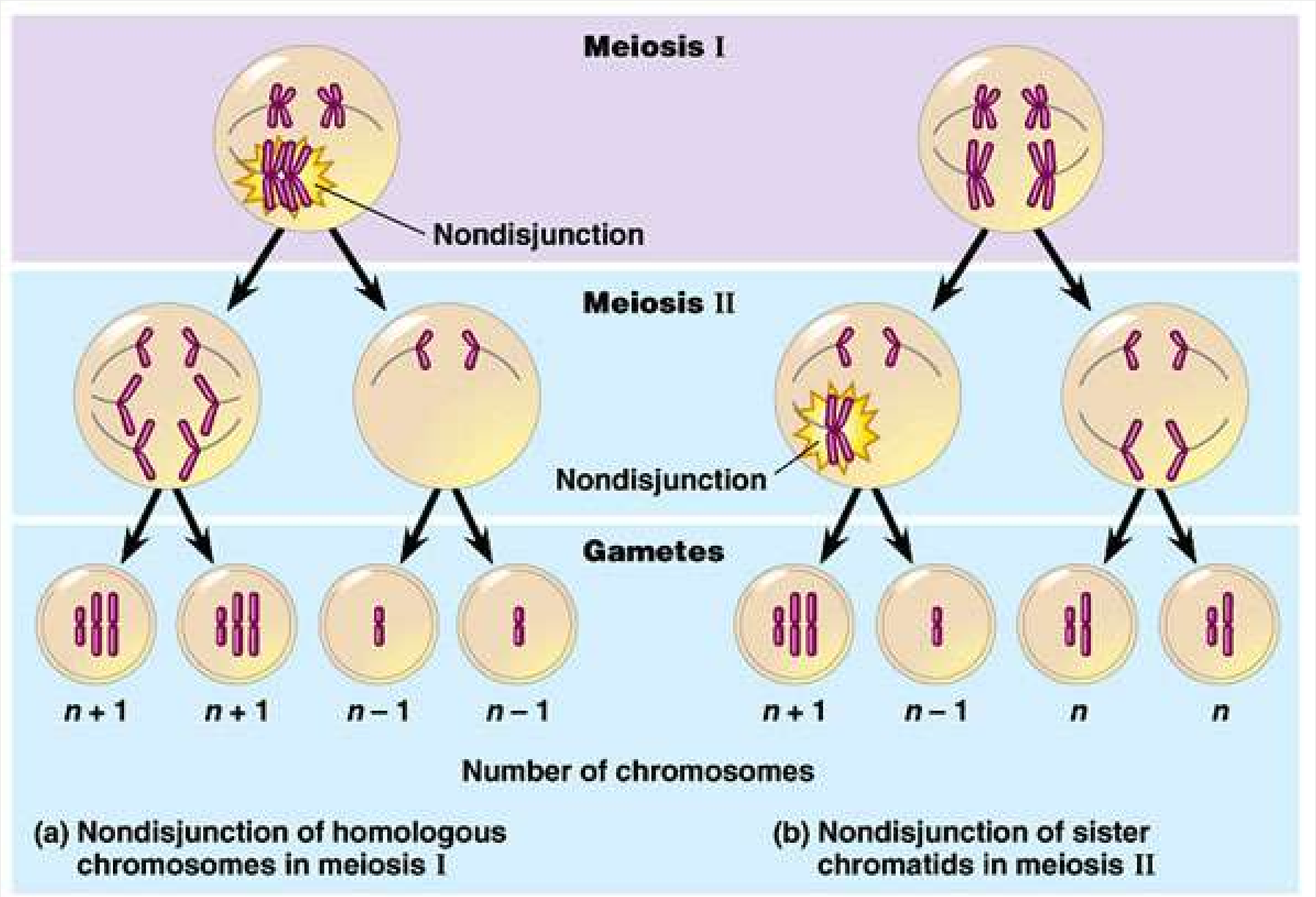
MISTAKES IN MEIOSIS:

- sometimes an accident occurs and the chromosomes fail to separate correctly
- this is called **NONDISJUNCTION**



NONDISJUNCTION

- recall: during meiosis I, one chromosome from each homologous pair moves to each pole of the cell…
- BUT, occasionally, both chromosomes of a homologous pair move to the SAME pole…the result:
- 2 types of gametes: one with an extra chromosome, one missing a chromosome



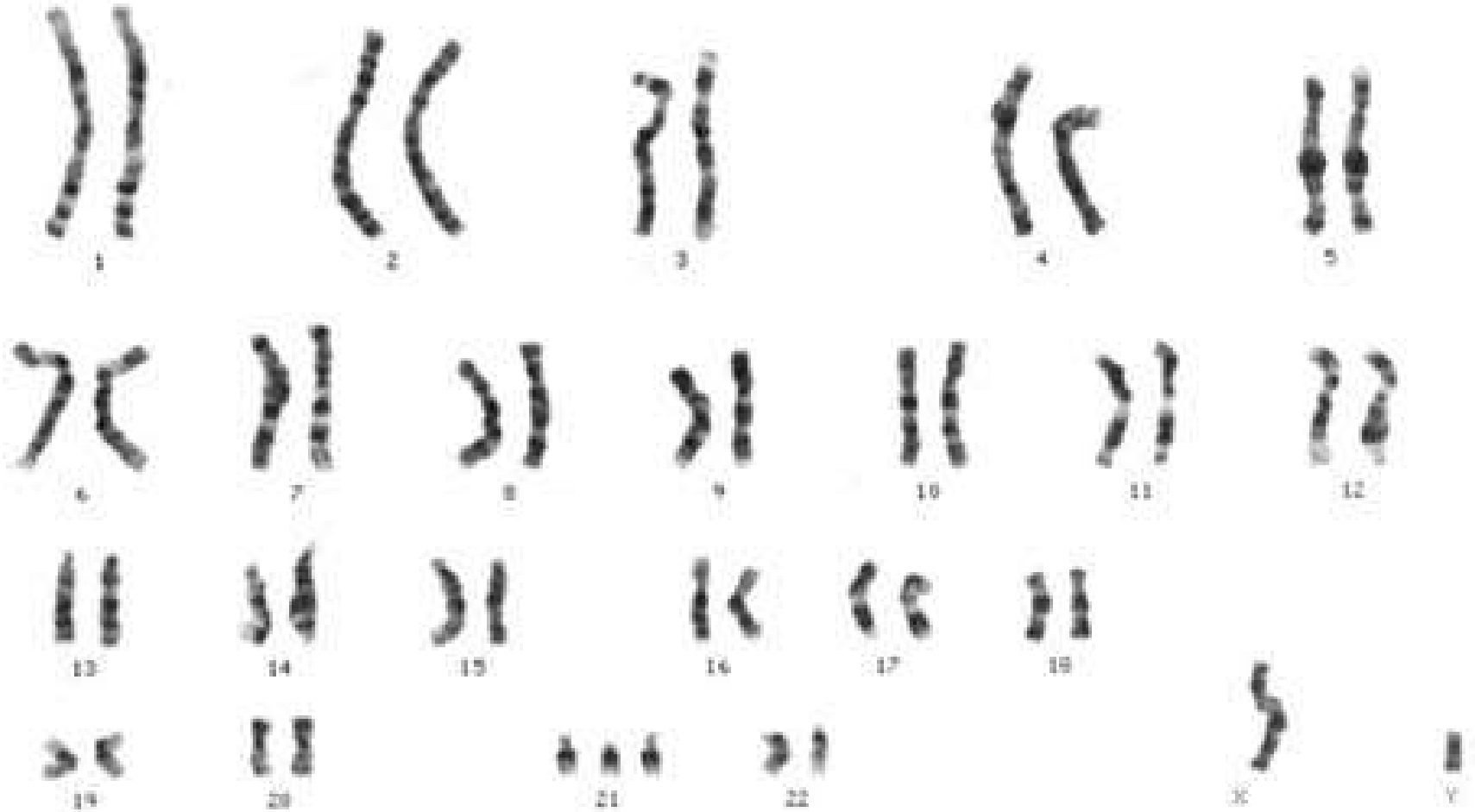
TRISOMY:

- a condition in which the **ZYGOTE** (the cell that is formed when a sperm and egg cell unite) has an extra copy of a chromosome

EXAMPLES:

➔ trisomy 21 (= Down Syndrome)

➔ XXY (= Klinefelter Syndrome)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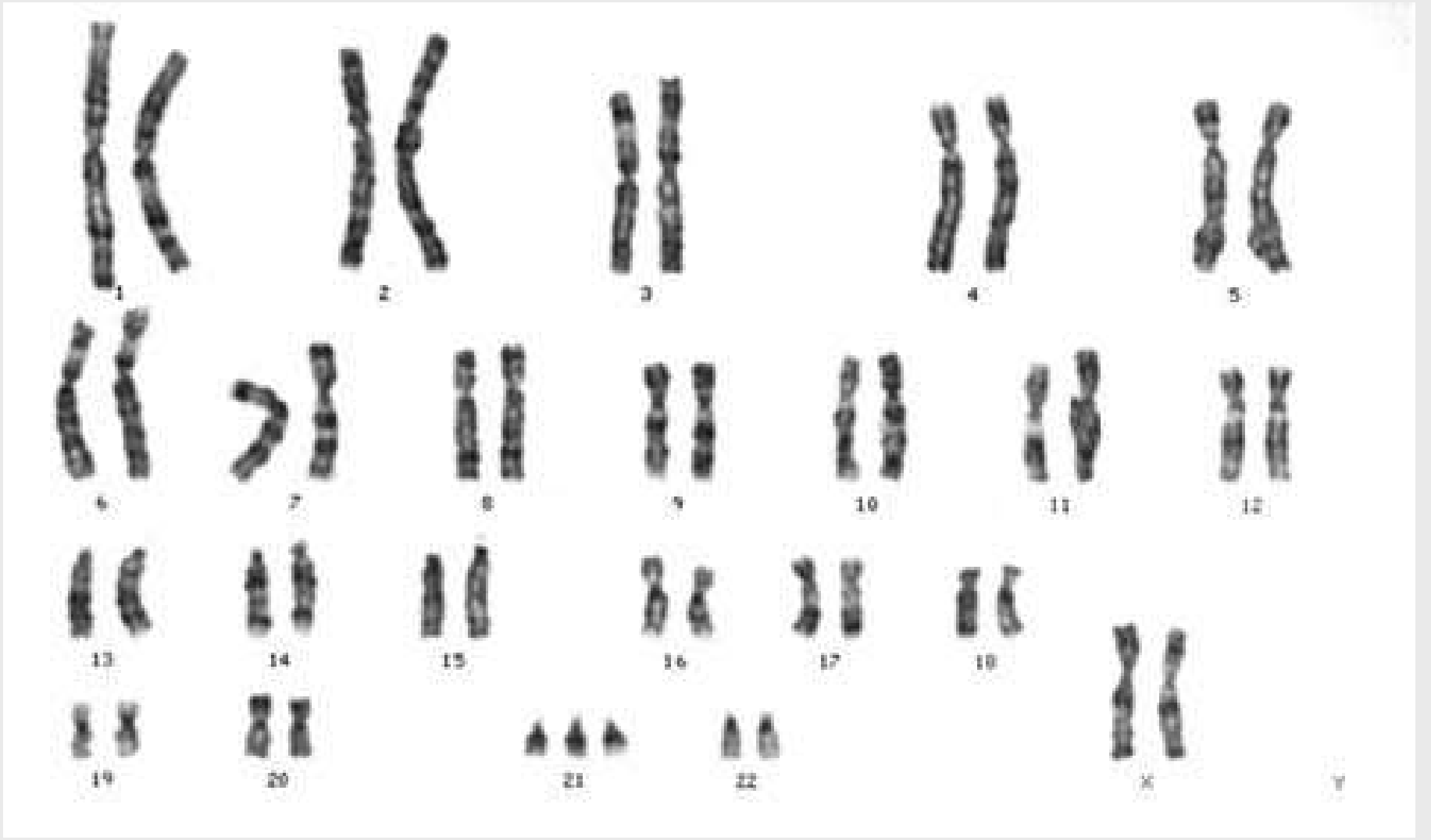
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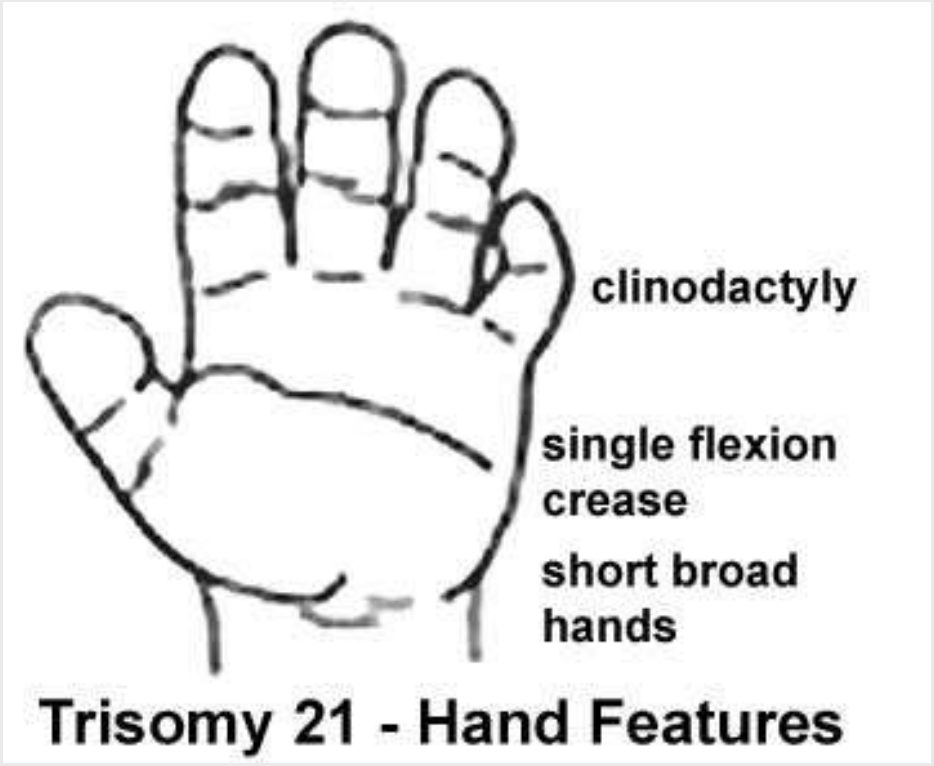
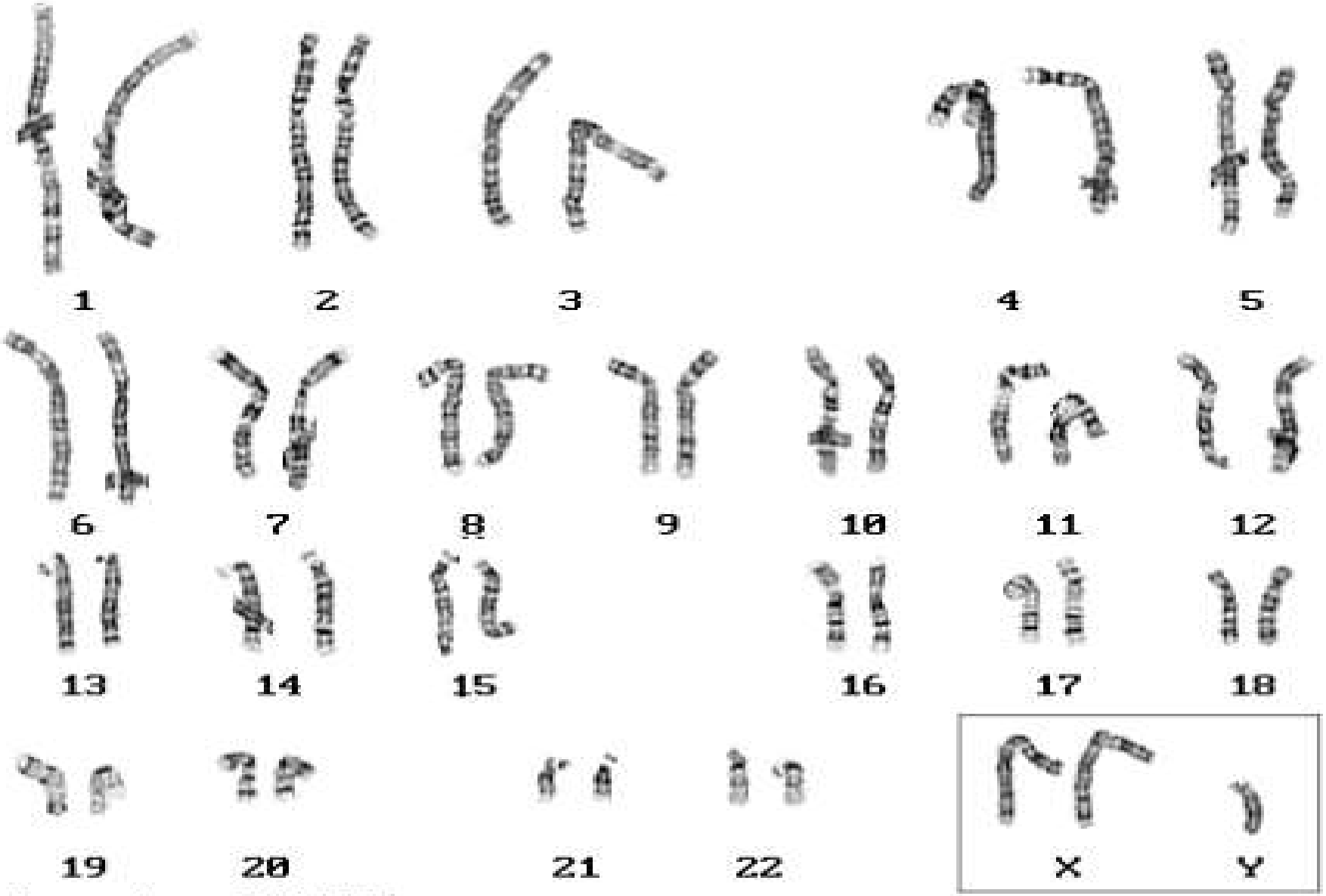

clinodactyly
single flexion crease
short broad hands
Trisomy 21 - Hand Features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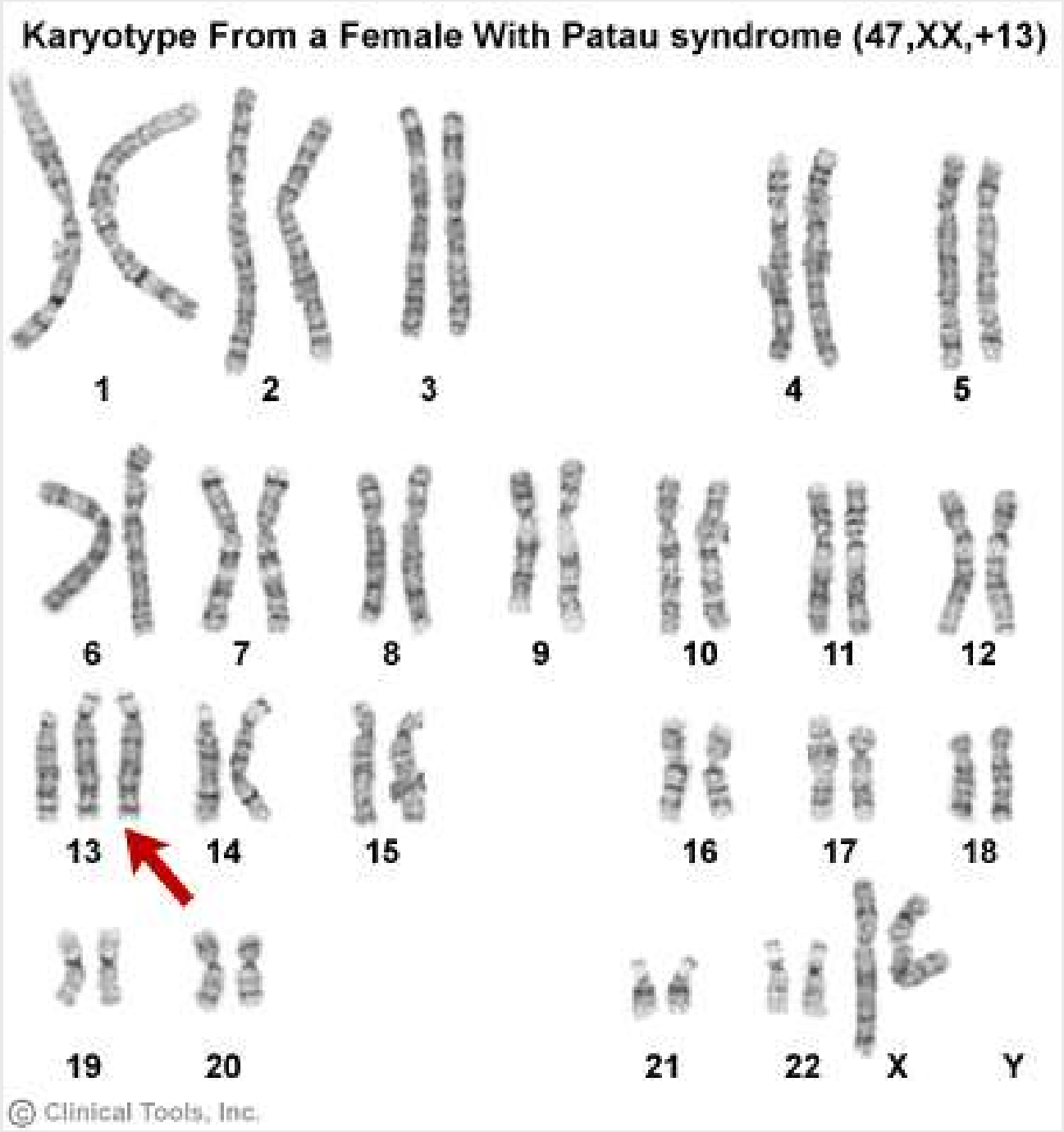
Karyotype: 47,XXY


Karyotype From a Female With Patau syndrome (47,XX,+13)
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Y
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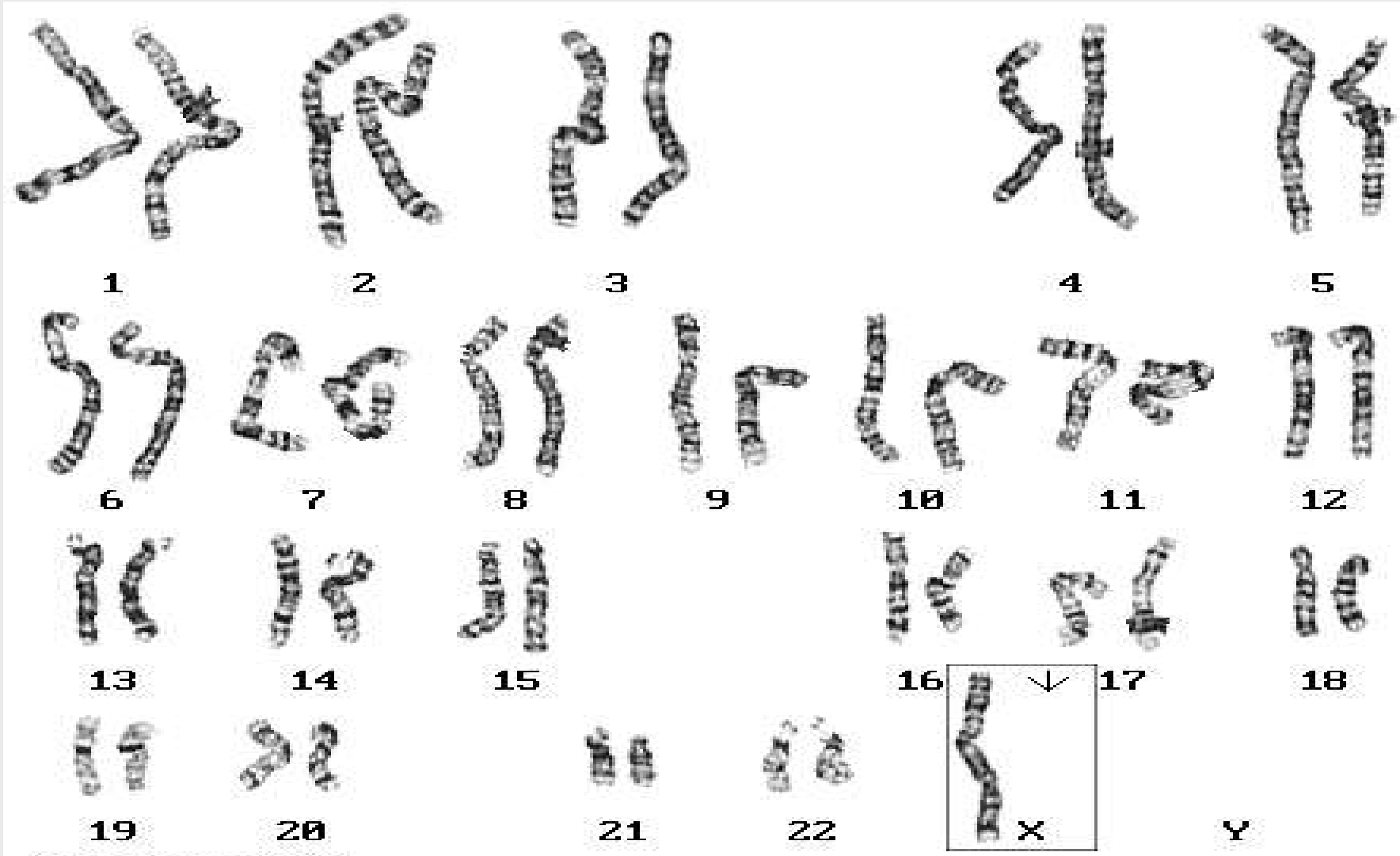
MONOSOMY:

- occurs when a gamete with a missing chromosome fuses with a normal gamete
- result is a zygote lacking a chromosome (only 1 copy = monosomy)
- most zygotes with monosomy do not survive

EXAMPLE of a non-lethal MONOSOMY:

➔ 1 X chromosome: Turner Syndrome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X
Y
Karyotype: 45,X

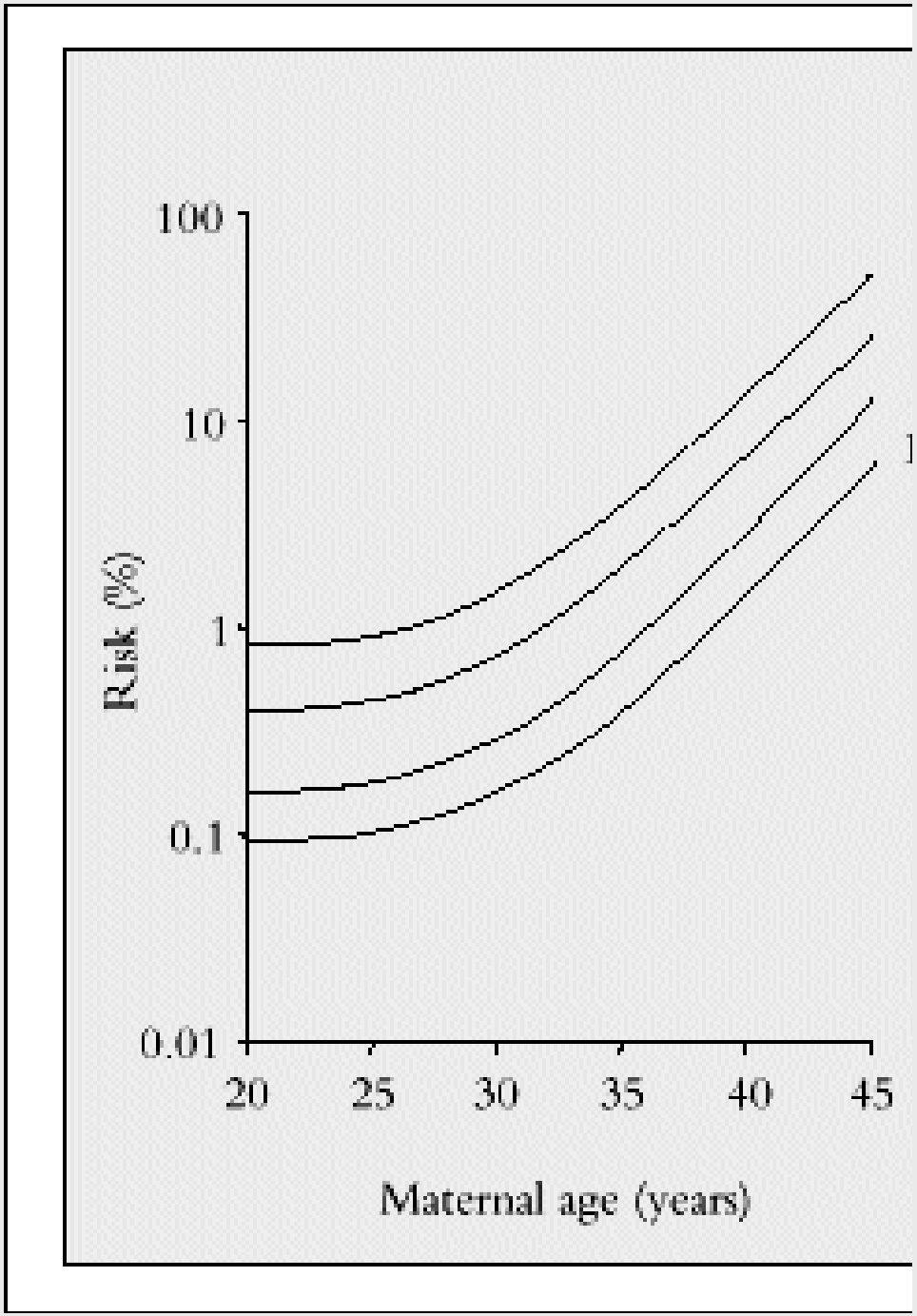
Who is at a greater risk for producing gametes that will result in trisomies or monosomies?

- keep in mind: females start meiosis before birth – their eggs reach prophase I and then halt…
- then, starting at puberty, one egg per month finishes meiosis…
- this lasts until she goes through a process called **menopause**, at which point egg production stops…SO…

Who is at a greater risk for producing gametes that will result in trisomies or monosomies?

- females who decide to become mothers at a later age

➔ Their egg cells have been in "halted" meiosis for a longer period of time, which means there is a greater chance that homologous chromosomes will "stick" together and fail to separate properly


100
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1
0.1
0.01
Risk (%)
20
25
30
35
40
45
Maternal age (years)

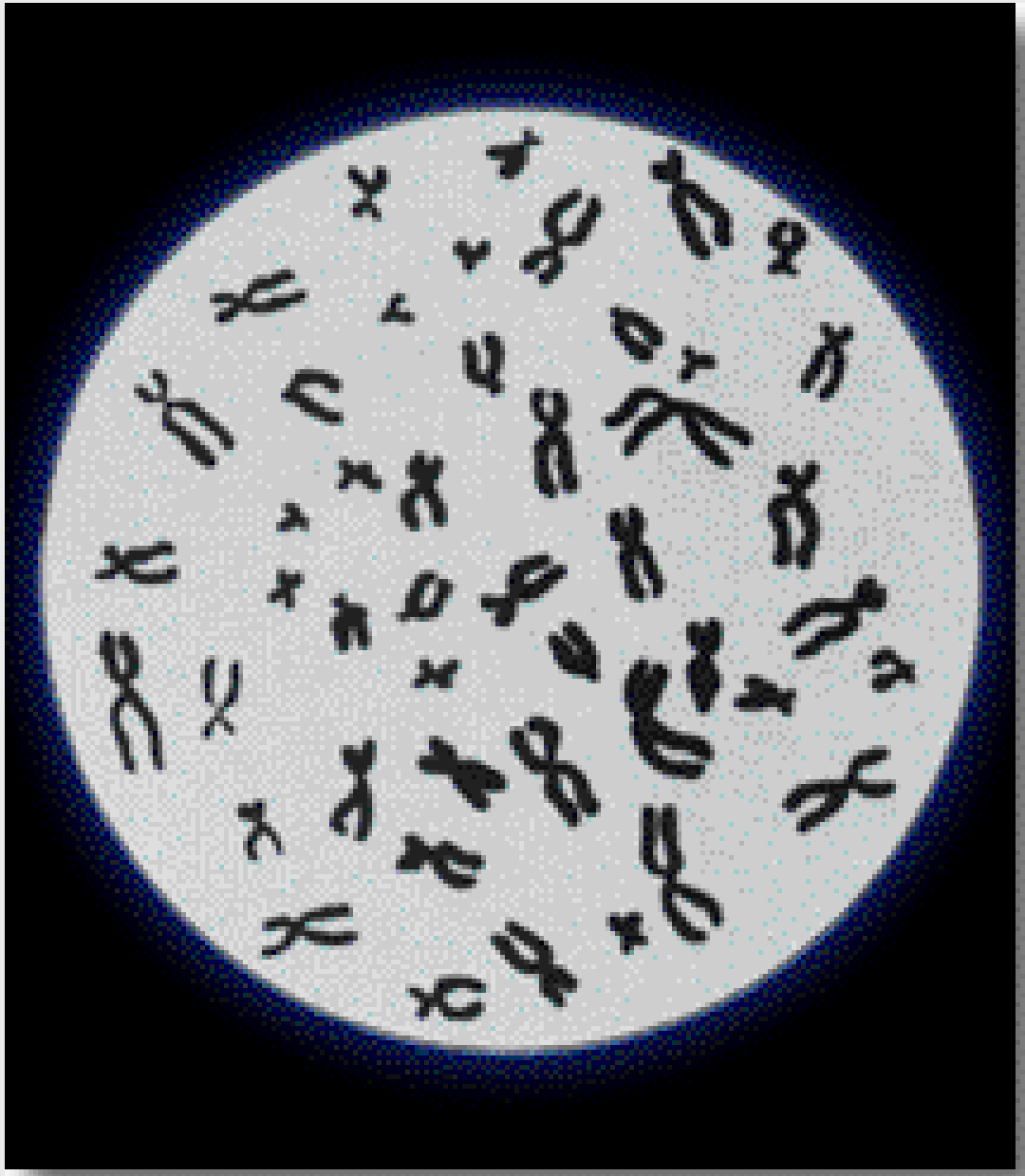
Can we test an unborn baby to see if they have an abnormal chromosome #?

- Yes!
- We can perform a test called a **KARYOTYPE**.

**__KARYOTYPE__: __a picture of someone's chromosomes__; can tell us:

➔ __sex__ (XX or XY?)

➔ if there are __extra__ or __missing chromosomes__

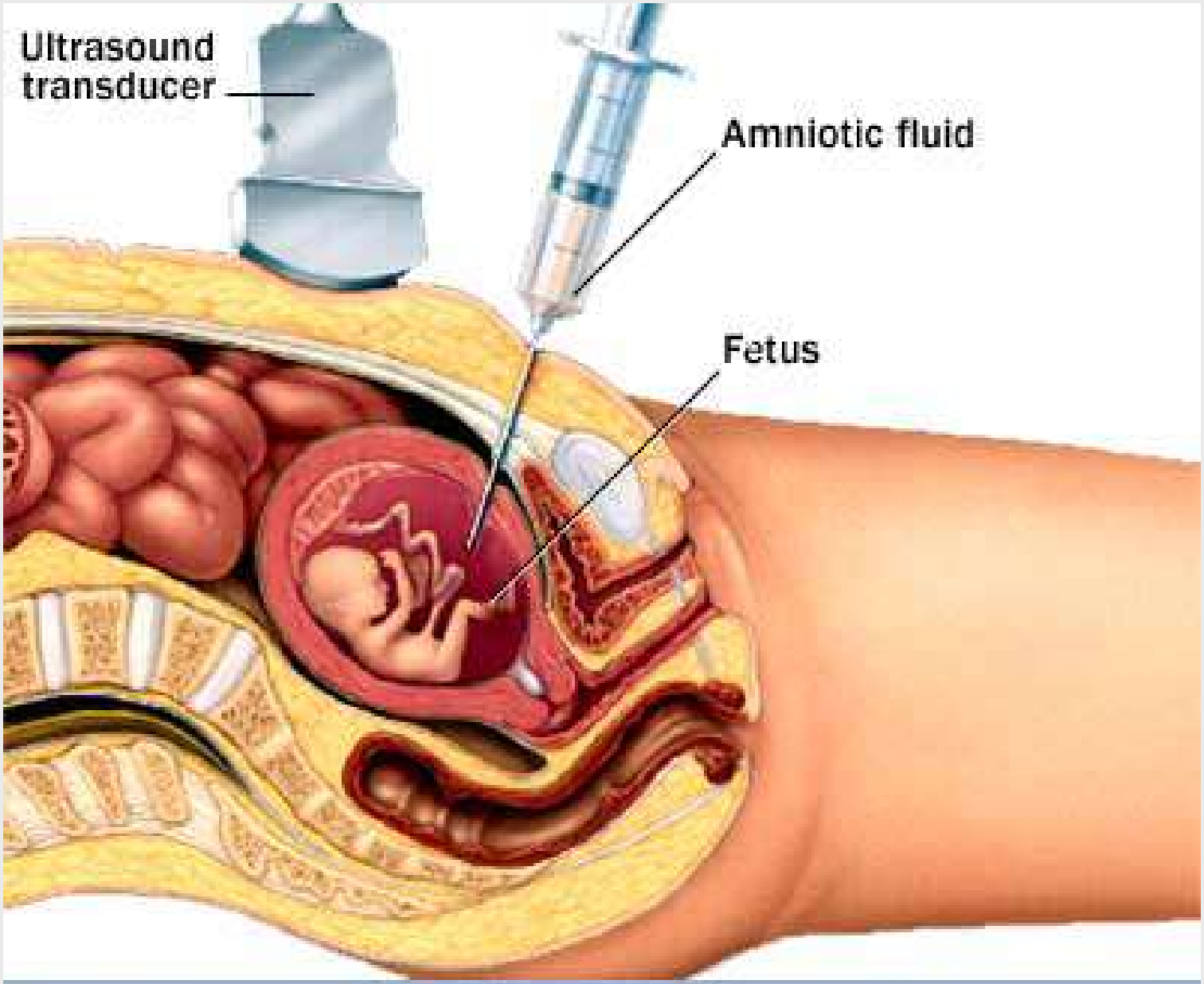


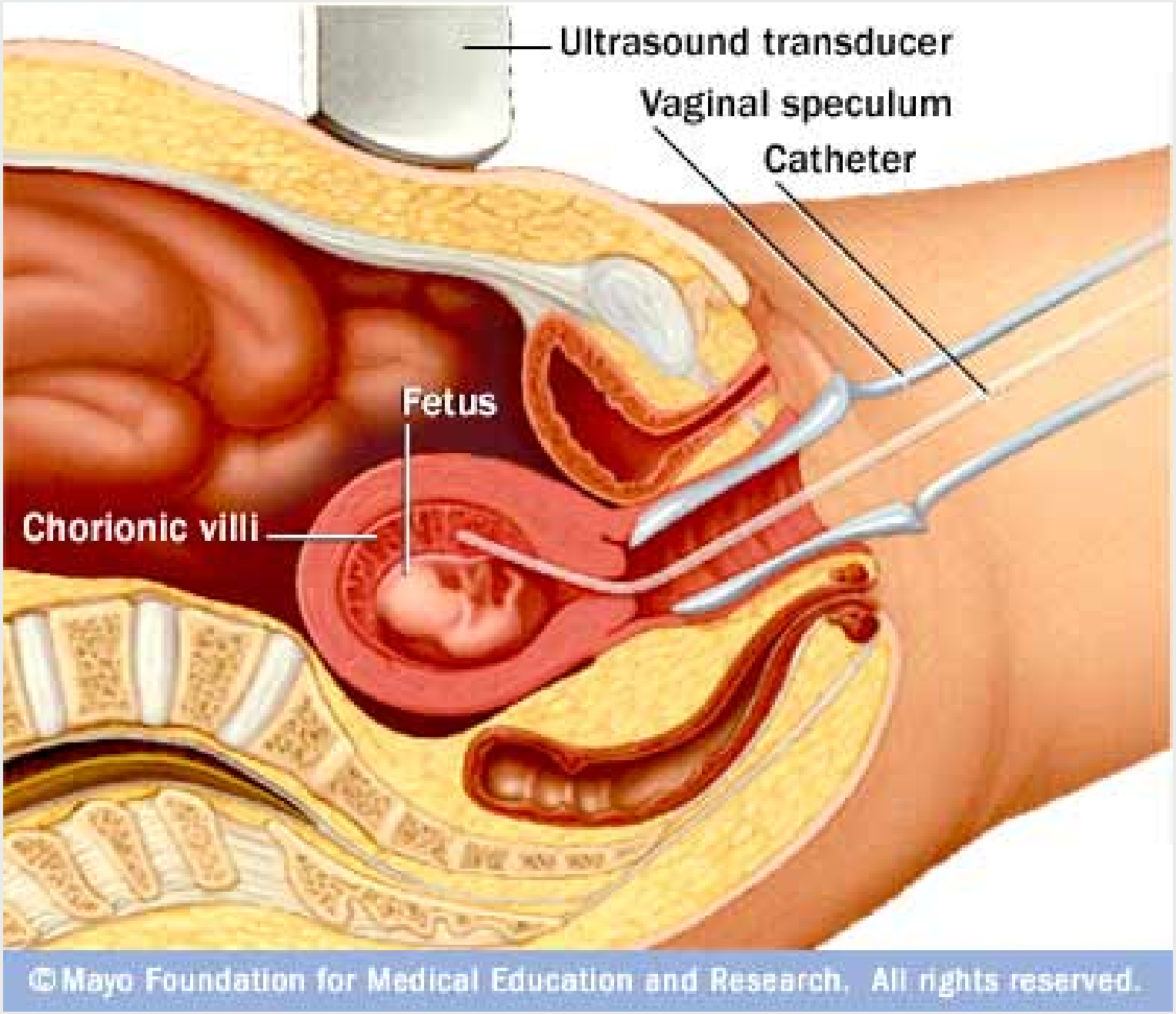
Steps Involved in a Karyotype:

1) Obtain fetal cells

➔ **amniocentesis** (BIG needle into mother's womb to gather amniotic fluid which contains fetal cells)

➔ **chorionic villus sampling** (remove cells directly from embryo's portion of placenta)





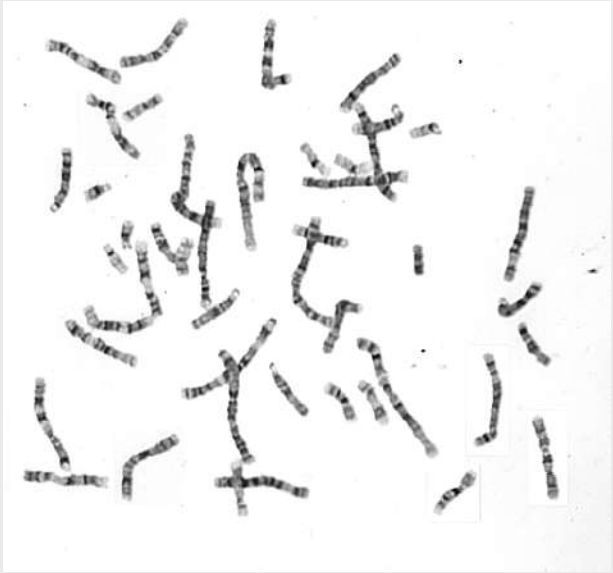
Steps Involved in a Karyotype:

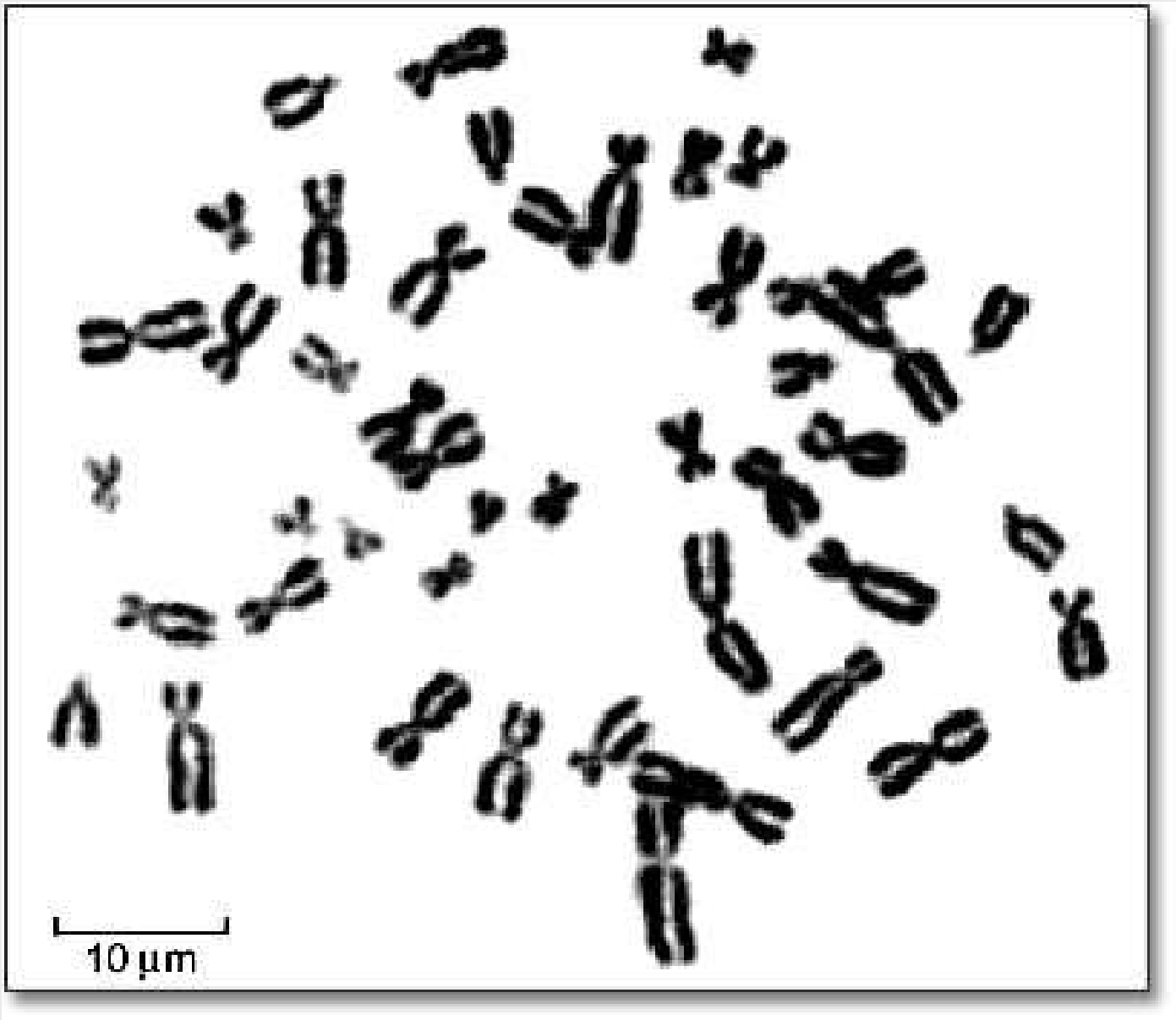
2) Grow cells in a petri dish ("in vitro") to increase their # and until they are in prophase of mitosis…WHY this phase and not interphase?

Steps Involved in a Karyotype:

3) Stop cell growth and break cells open to release chromosomes

4) Prepare a chromosome "spread" on a microscope slide



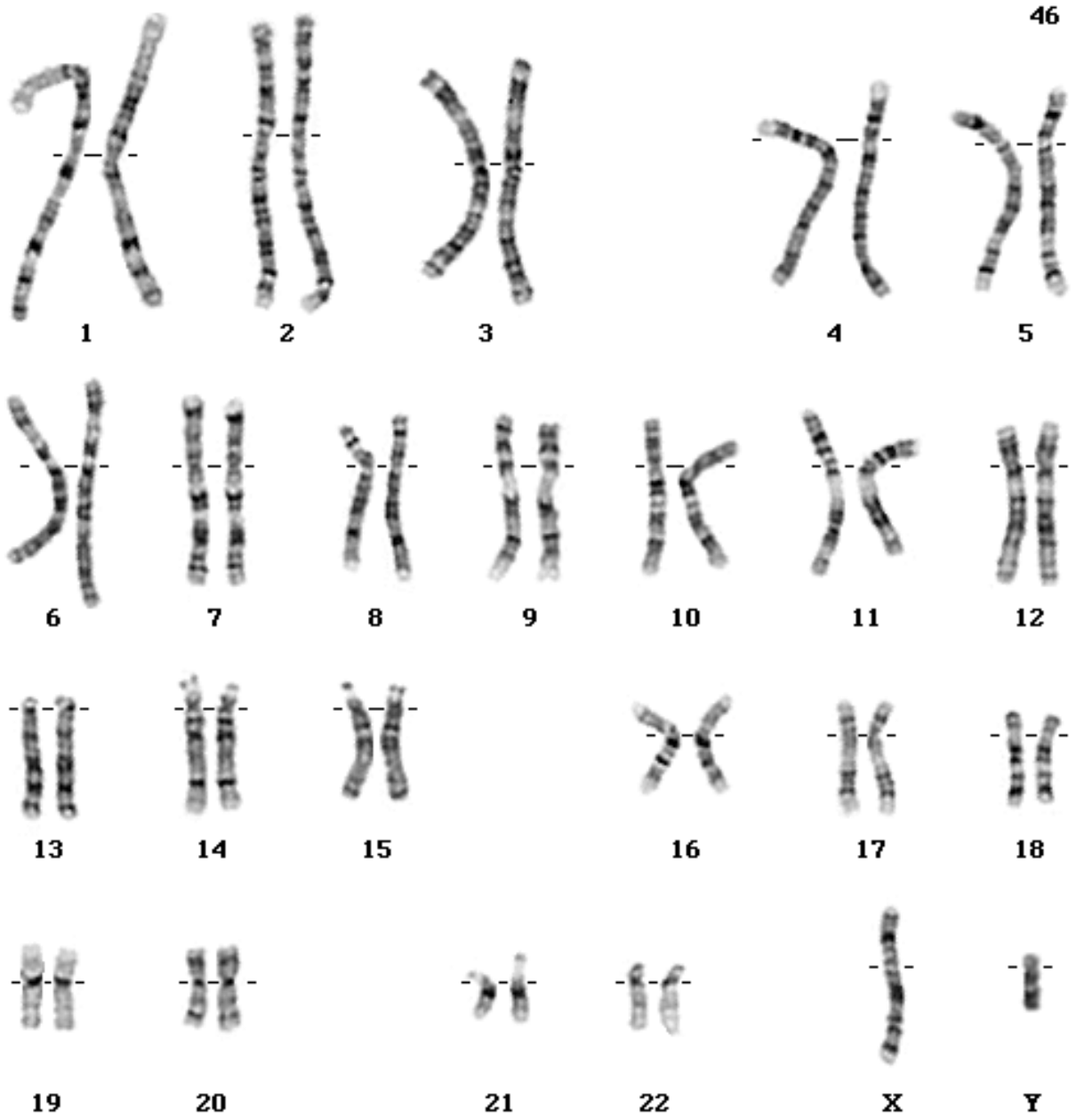

10 µm

Steps Involved in a Karyotype:

5) Photograph and sort chromosomes to determine:

➔ gender

➔ if # of chromosomes is correct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A Karyotype CANNOT tell us:

-if there are any mutations in genes (i.e. point mutations, frameshift mutations, etc.)

-if there are genetic disorders such as cystic fibrosis, sickle cell anemia, etc.

A Karyotype CANNOT tell us:

-whether they have a dominant or recessive phenotype

-who the parents are

-if they will be as smart as Mr. Davies