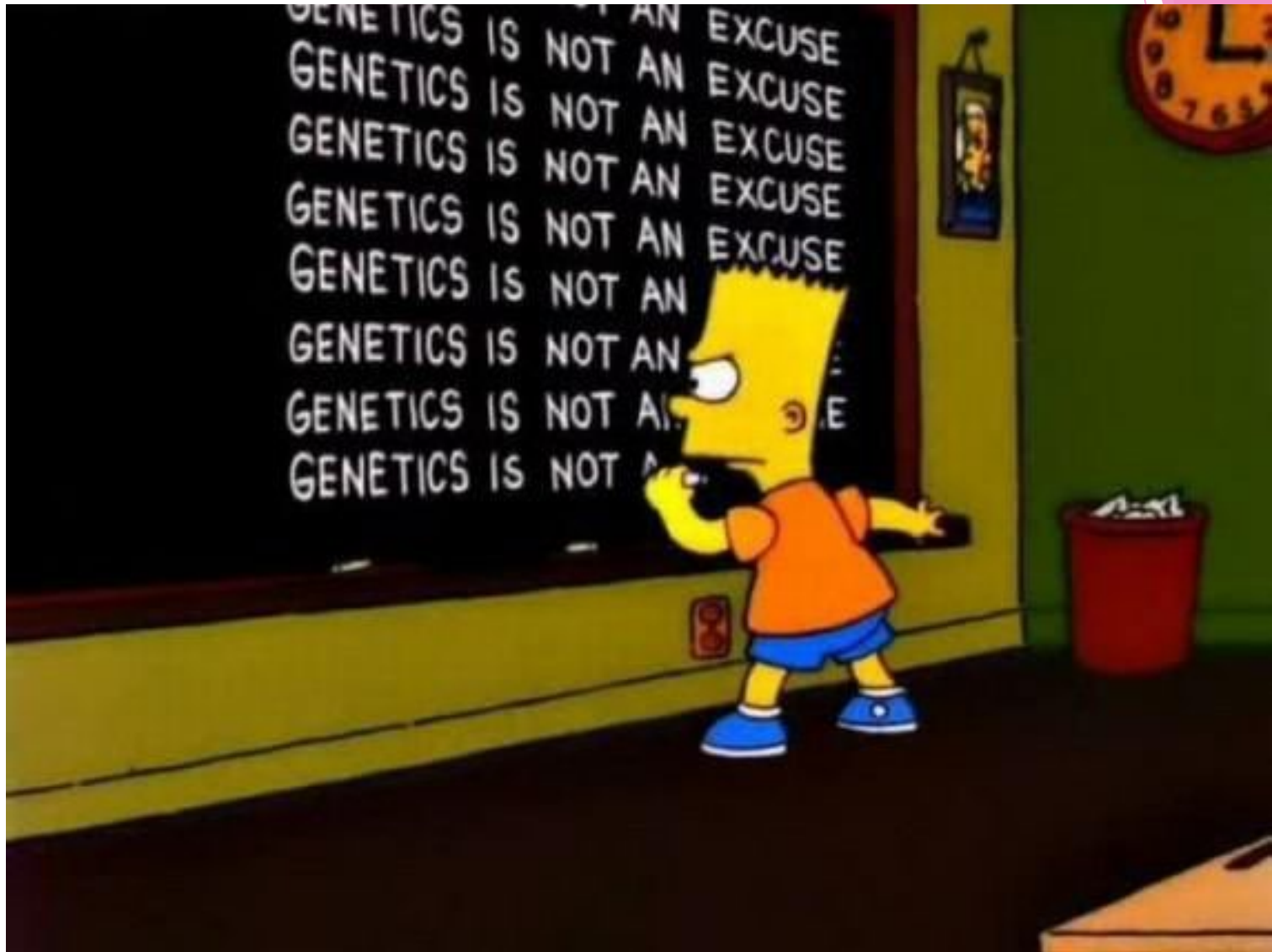


Genetics

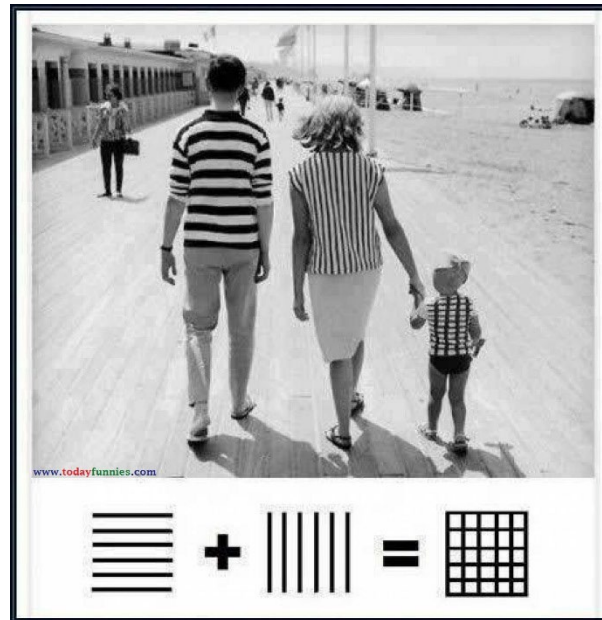


Curriculum

- ▶ describe, in general, the behavior of chromosomes during mitosis, meiosis and fertilization
- ▶ describe the role of proteins in the human body as regulatory molecules (enzymes), as structural molecules and as a source of energy

Genetics

- ▶ The study of heredity and variation in organisms
- ▶ Genetics is the reason we look and act the way that we do
- ▶ Everything we do is based on information found in the nuclei of our cells



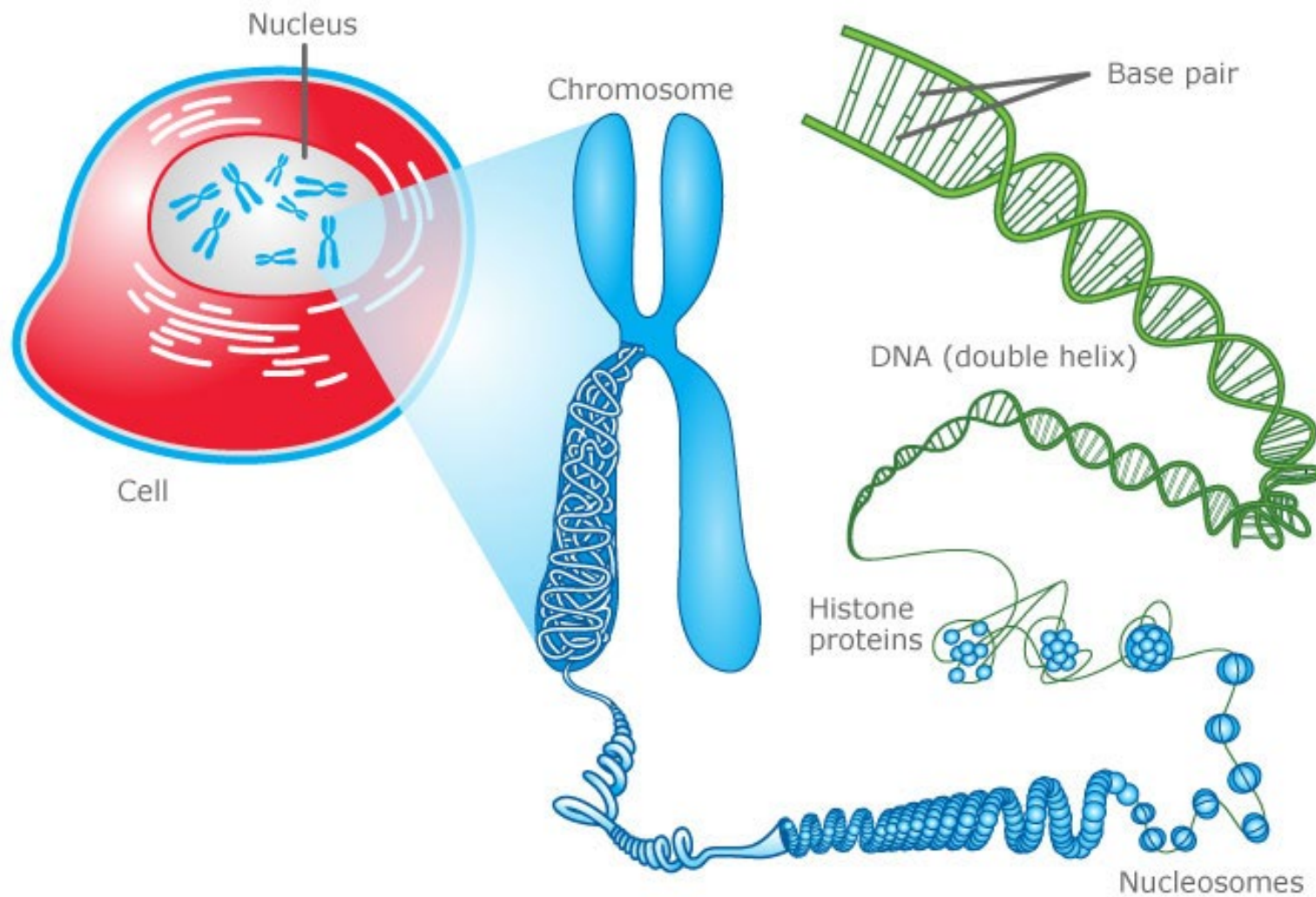
Chromosomes

- ▶ **Chromosomes** are long X shaped strands of DNA that act as an instruction booklet to make us
- ▶ Chromosomes are long thin strands of DNA coiled around protein molecules for protection
- Humans have 23 pairs of chromosomes
- Each species has its own number of chromosomes

Diploid Chromosome Number

Goldfish	
Potato	
Human	
Pea	
Fruit fly	

Chromosomes



Chromosomes

- ▶ Chromosomes come in pairs with half being used, and half as a backup.
- ▶ We receive half of our chromosomes from our mom and half from our dad, so each half of our chromosomes, although almost identical, have slight differences
- ▶ Each cell has all 23 pairs of chromosomes, but only uses the specific information it needs

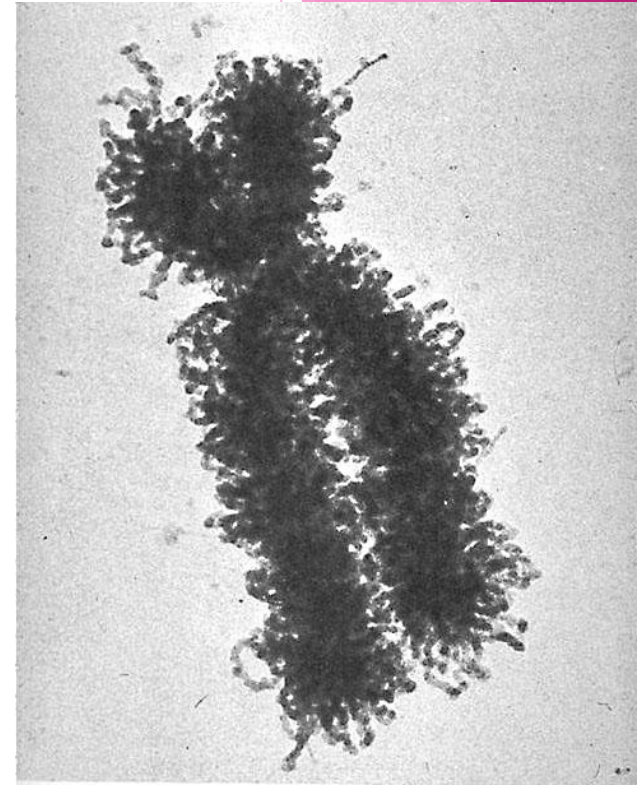
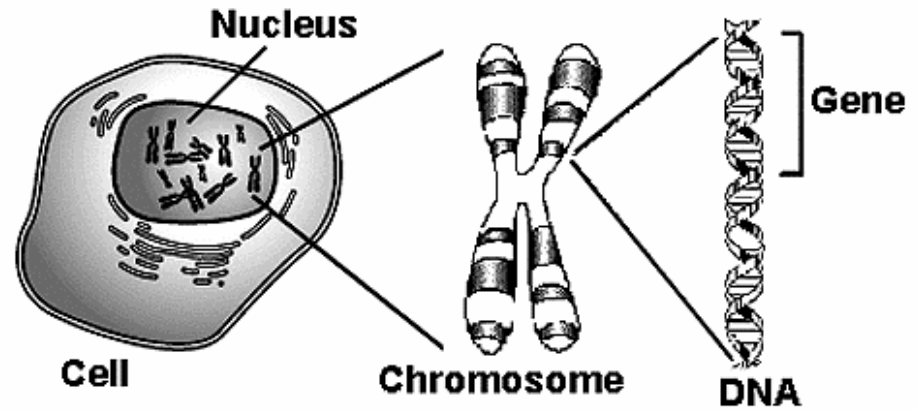


FIGURE 1-14
An electron micrograph of a human chromosome.
Chromosome XII from a HeLa cell culture. (Courtesy
of Dr. E. Du Praw.)

Genes

- ▶ A **gene** is an uninterrupted segment of deoxyribonucleic acid, DNA, which contains coded instructions
- ▶ Genes come in pairs
- ▶ Each pair of genes carries DNA for the same trait (for example, eye color)
- ▶ These pairs of genes are always found at the same position on a chromosome
- ▶ However, the code for each gene in the pair may be different



The Role of Proteins

- ▶ The information coded in DNA, tells the body which proteins to make with that information
- ▶ If all the water was removed from the body, 50% of the dry mass leftover would be proteins

Type of Protein	Role of Protein
Enzymes	Speed up chemical reactions in the body
Hormones	Act as signals to coordinate and regulate activities in the body
Structural	Support cells and provide frameworks for other proteins to attach to
Transport	Allow the movement of materials within cells or the body (for example, hemoglobin)
Defensive	Protect the body from pathogens (for example, antibodies)
Energy	Decomposition of certain proteins can serve as a source of energy

Human Karyotype

- ▶ Chromosomes are not neatly organized in the nucleus and look like a plate of spaghetti
- ▶ A **karyotype** is an organized version of chromosomes that have been matched up into their respective pairs called **homologous chromosomes** and arranged from the largest pair to the smallest pair
- ▶ Geneticists use 3 criteria to identify and match-up chromosomes:
 - ▶ Length of chromosome
 - ▶ Pattern of dark bands produced on each chromosome
 - ▶ Position of chromosome's constricted part, called **Centromere**

Board Question

Which chromosome is the best match for Chromosome 1?



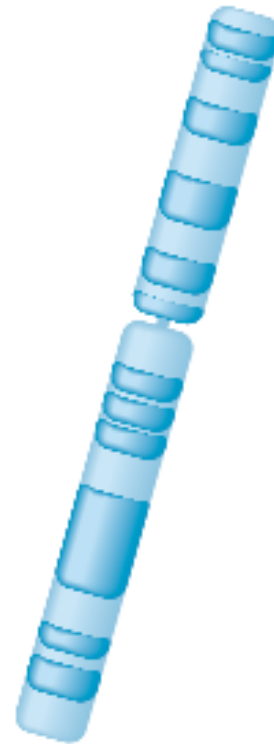
A



B

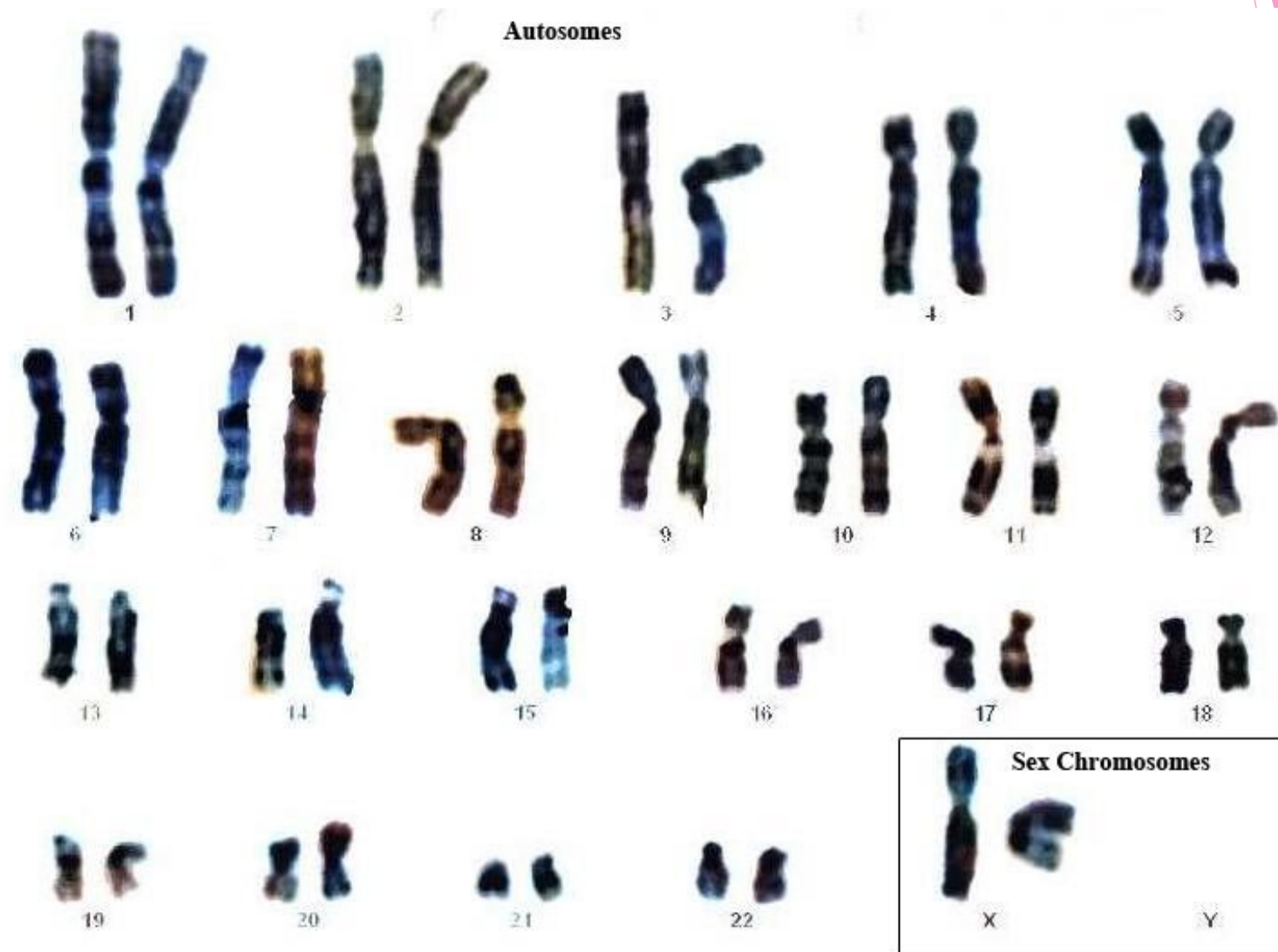


C



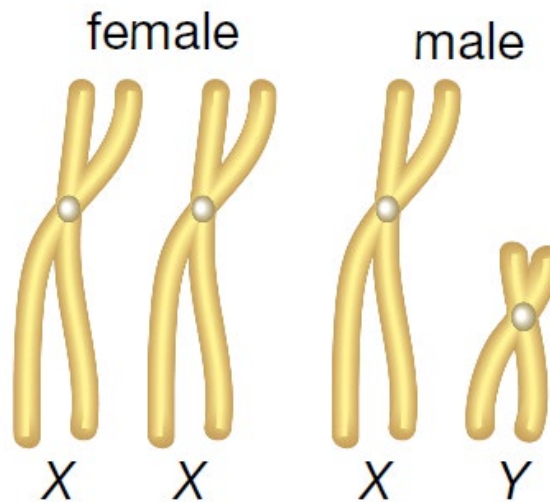
Chromosome 1

Real life karyotypes



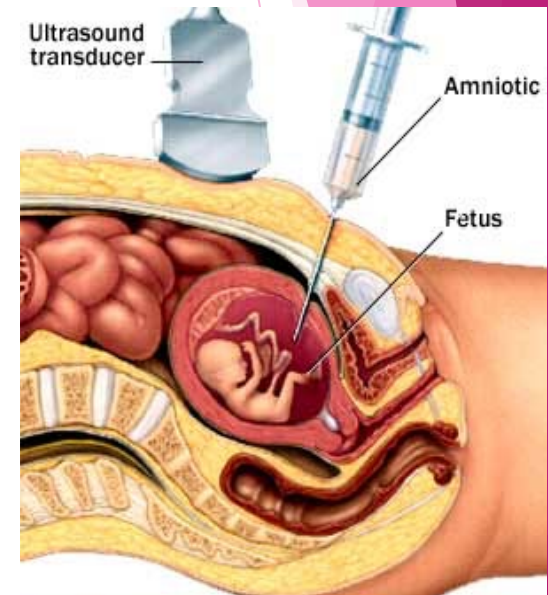
Sex Chromosomes

- ▶ The last two chromosomes are **sex chromosomes** and determine the gender of an organism
- ▶ If an individual has two X chromosomes, she is a female
- ▶ If an organism has an X and a Y, then he is a male.



Amniocentesis

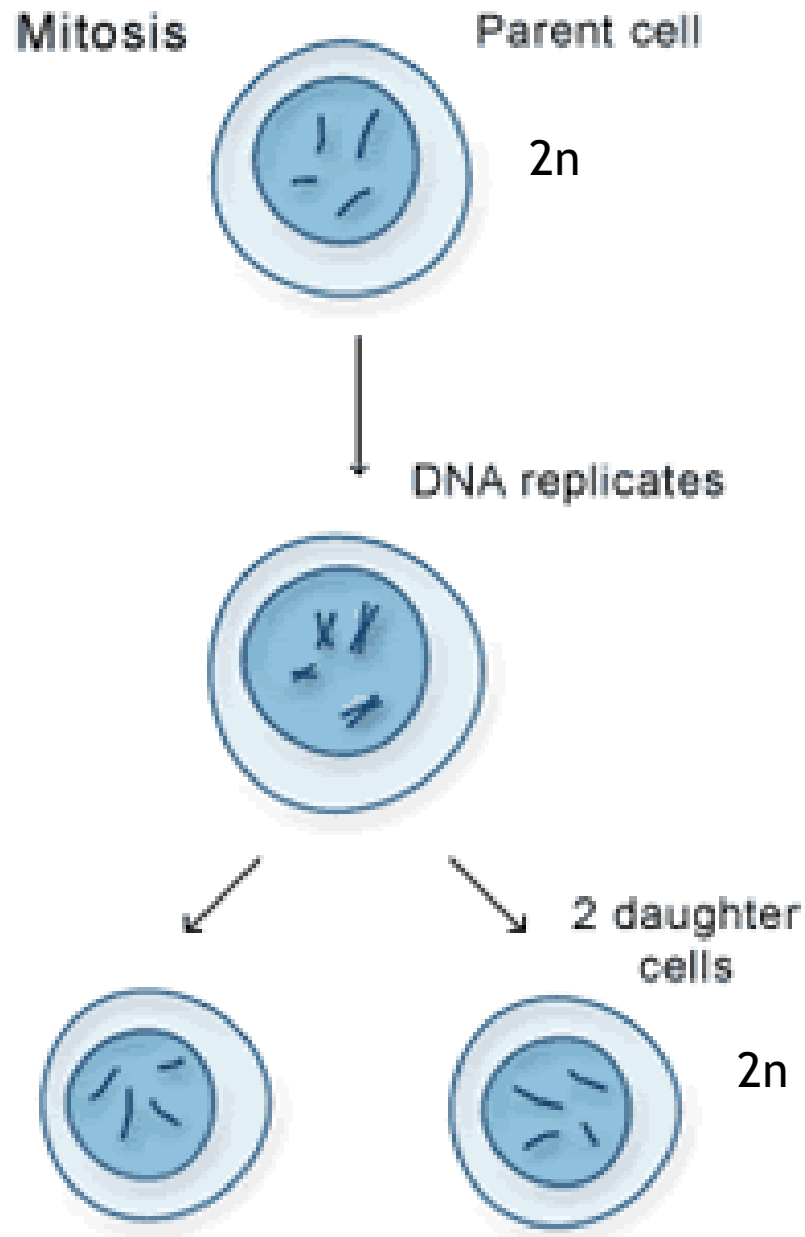
- ▶ **Amniocentesis** is a prenatal test done to check the karyotype of an unborn child
- ▶ Using an ultrasound transducer, a long needle draws amniotic fluid outside of the fetal womb
- ▶ The result is an analysis of the fluid to check for abnormalities (ie. Down Syndrome) in the fetus
- ▶ It can lead to increased risk of miscarriage



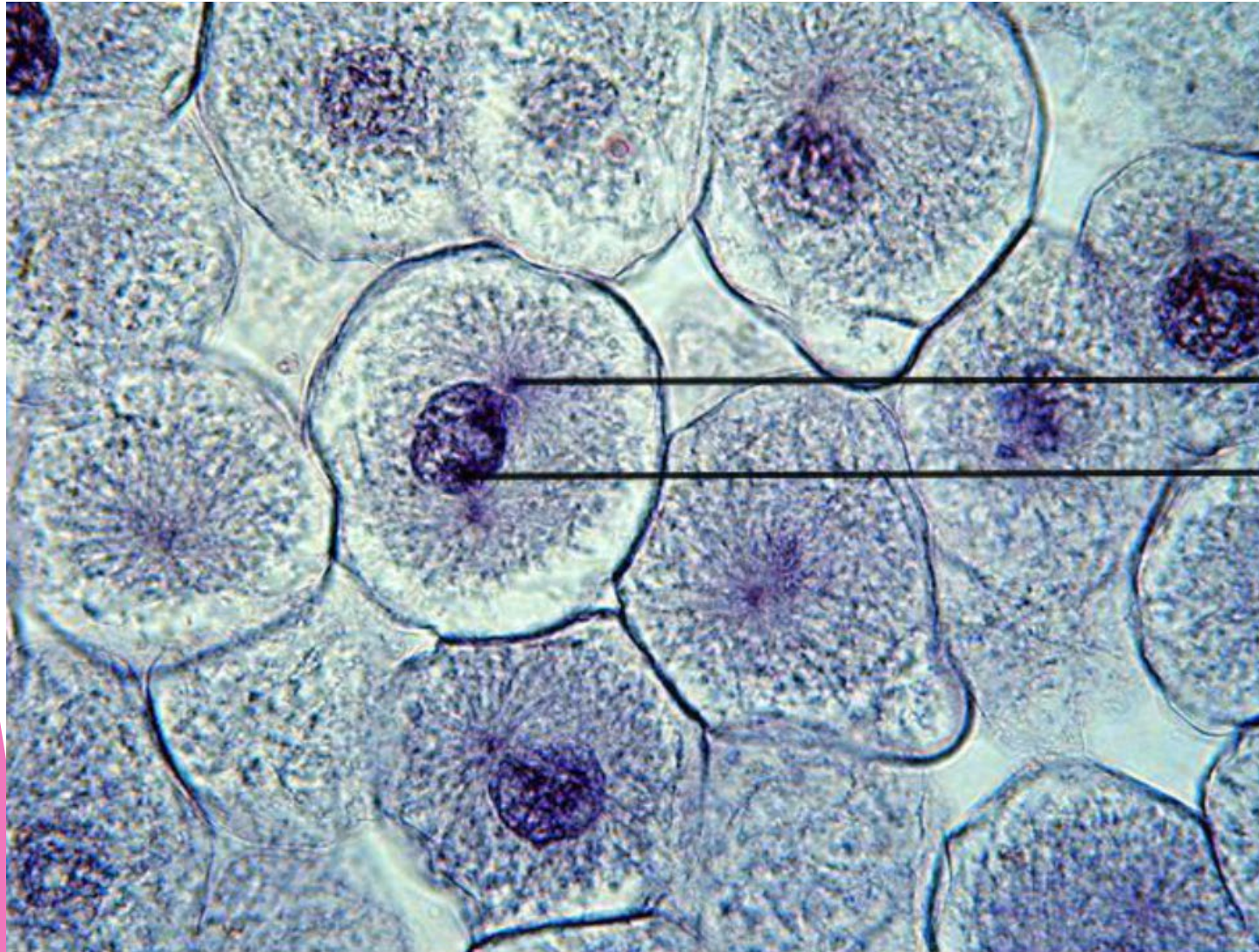
Mitosis ($2n$)

- ▶ You are asexually reproducing as we speak
- ▶ Old cells die, new cells need replace them. These cells in your body are called **autosomal cells**
- ▶ They do this by growing larger and splitting in a process called **mitosis**
- ▶ Before splitting they replicate their DNA so the new cell has 23 pairs of chromosomes as well
- ▶ The new cells created are identical and called **daughter cells**
- ▶ The daughter cell and the original cell are called **diploid cells** because each has 2 copies of each chromosome type

Mitosis



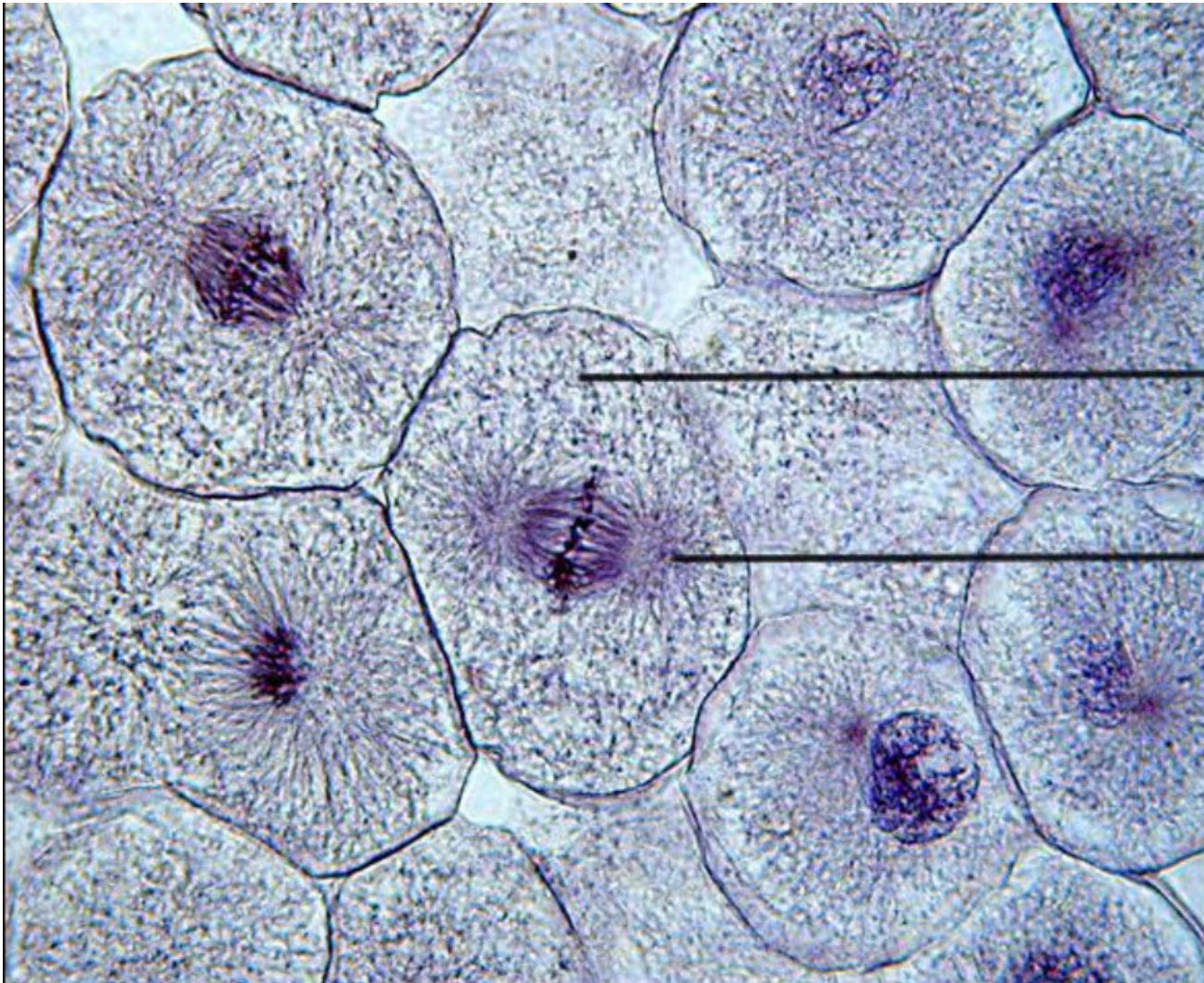
Prophase of mitosis



Centriole

Note:
chromosomes
are visible

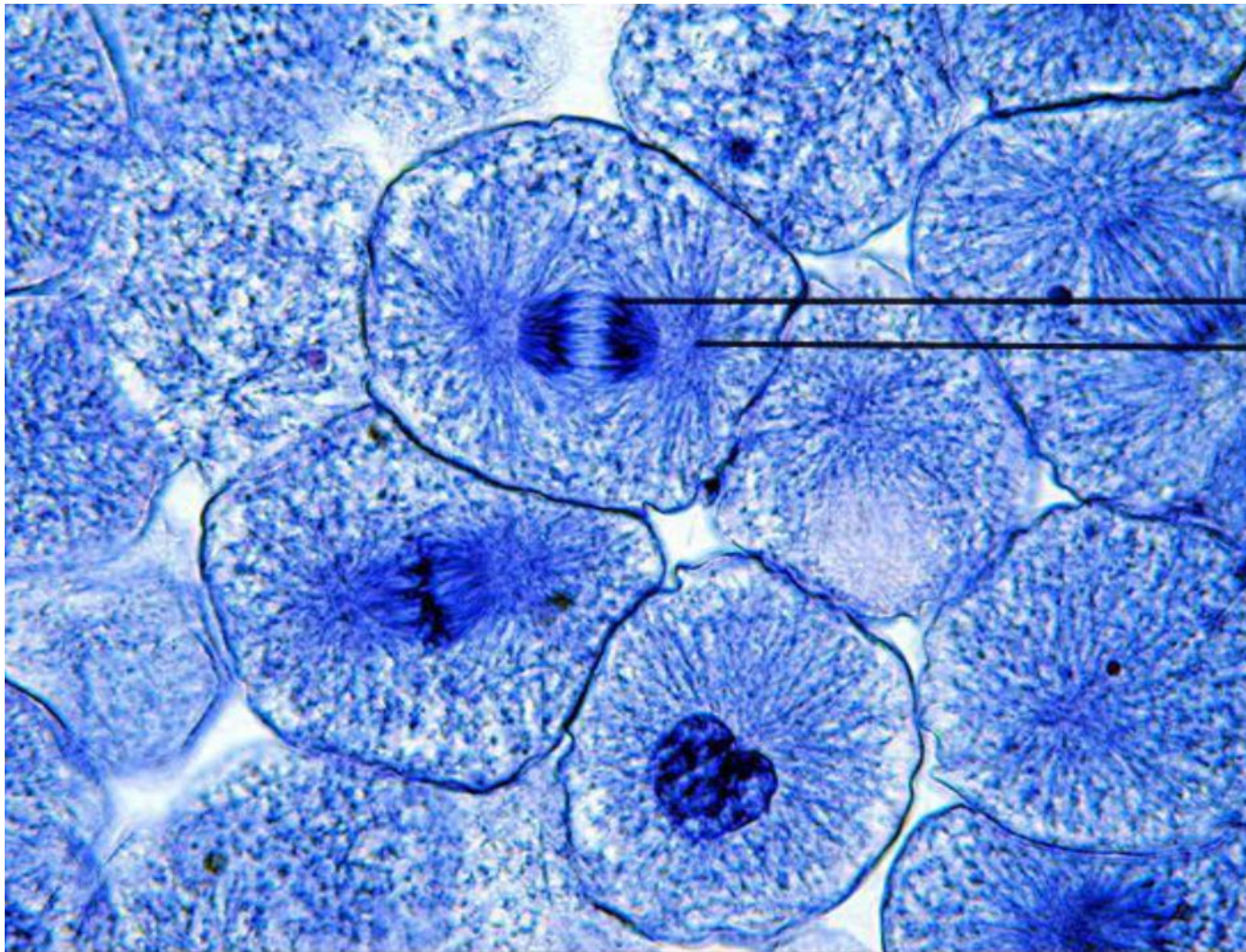
metaphase



Cell in metaphase

Centriole

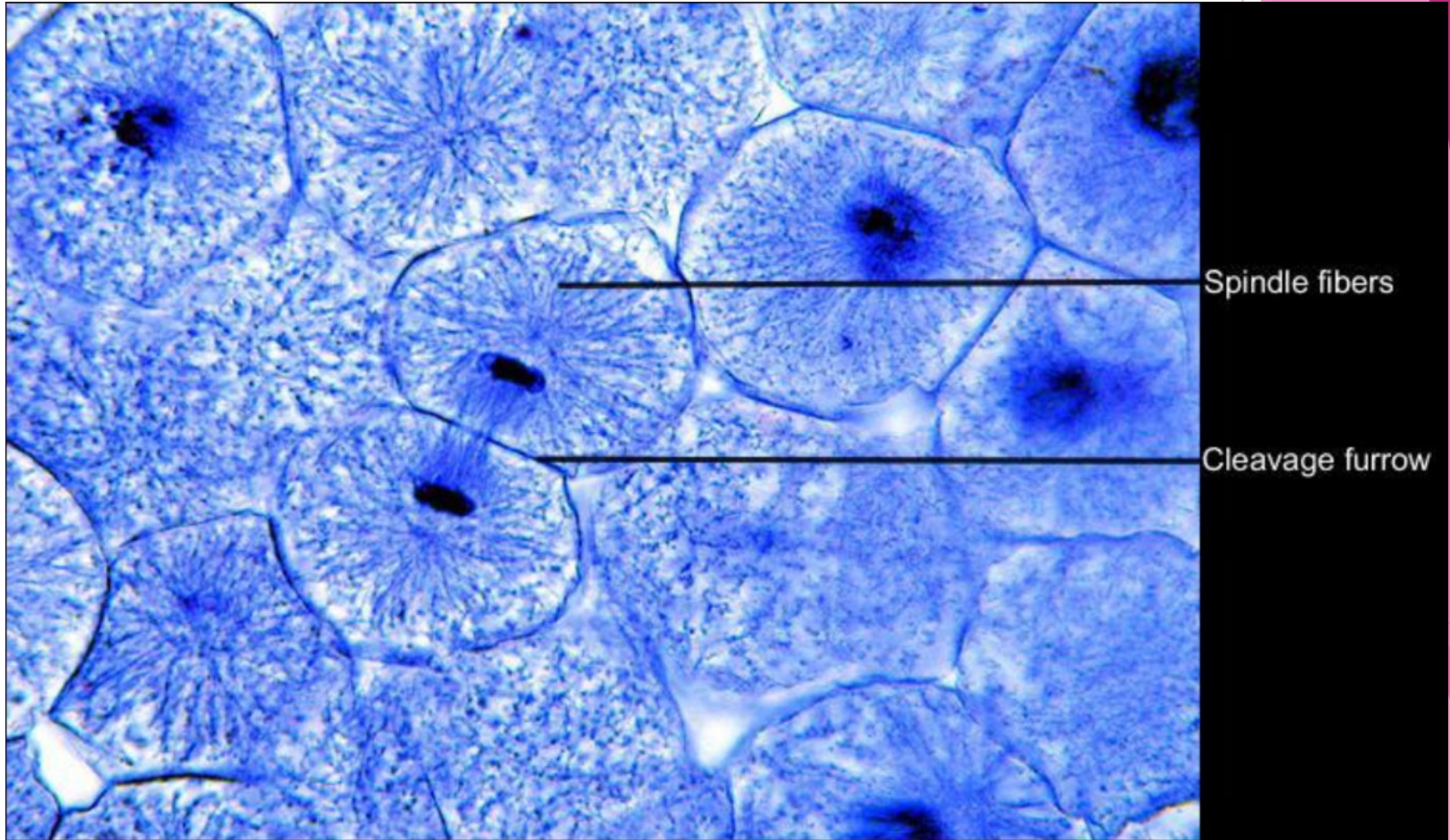
Anaphase



Chromosomes
Centriole

Anaphase of Mitosis (1000x)

telophase



Telophase of Mitosis (1000)

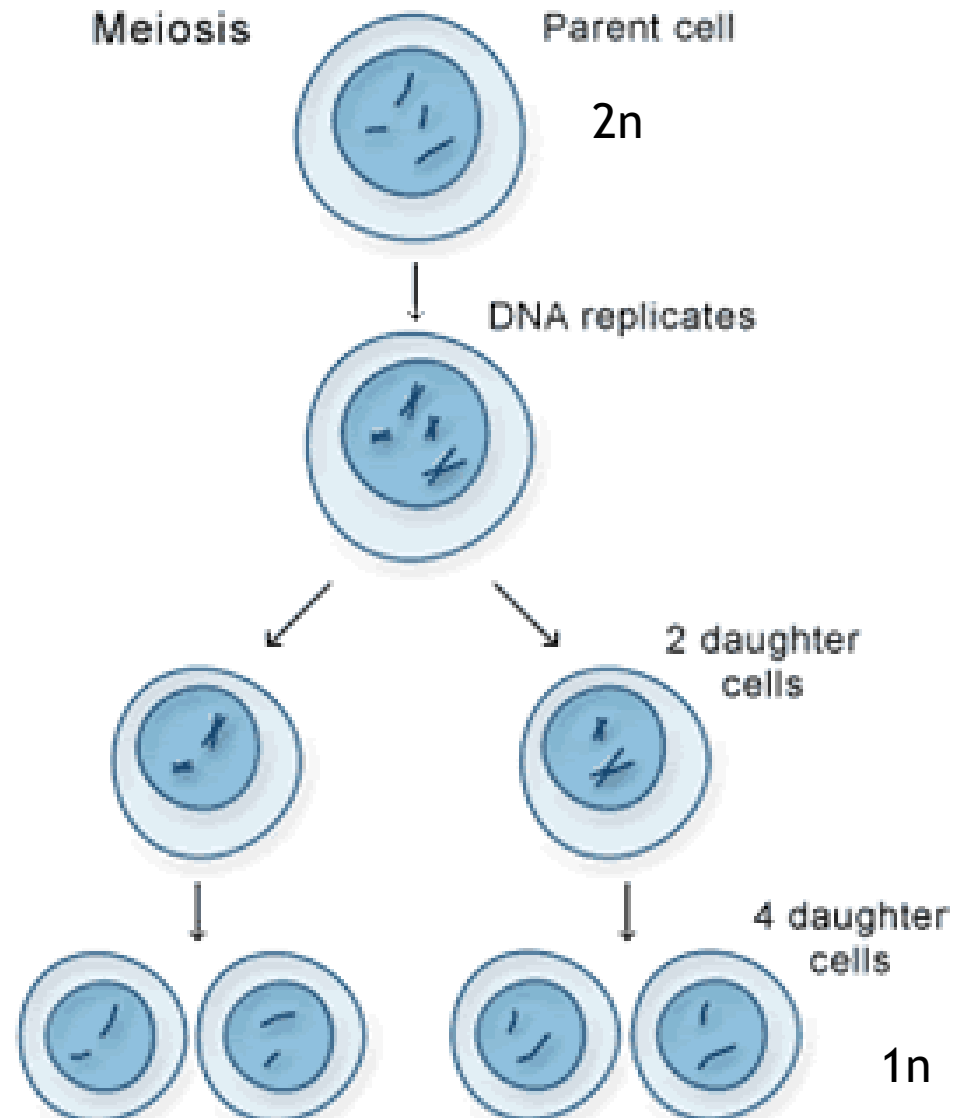
Videos mitosis (8 min)



Meiosis (n)

- ▶ Sexual reproduction is a little more difficult and requires sperm and eggs called **gametes**
- ▶ The main differences are during meiosis:
 - ▶ when homologous chromosomes meet up, they swap DNA with each other in a process called **crossing over**
 - ▶ During the process two divisions occur
 - ▶ When the gametes are made in the end, they have half the chromosomes called a **haploid cell** compared to a normal autosomal cell

Meiosis



Videos (8 min)

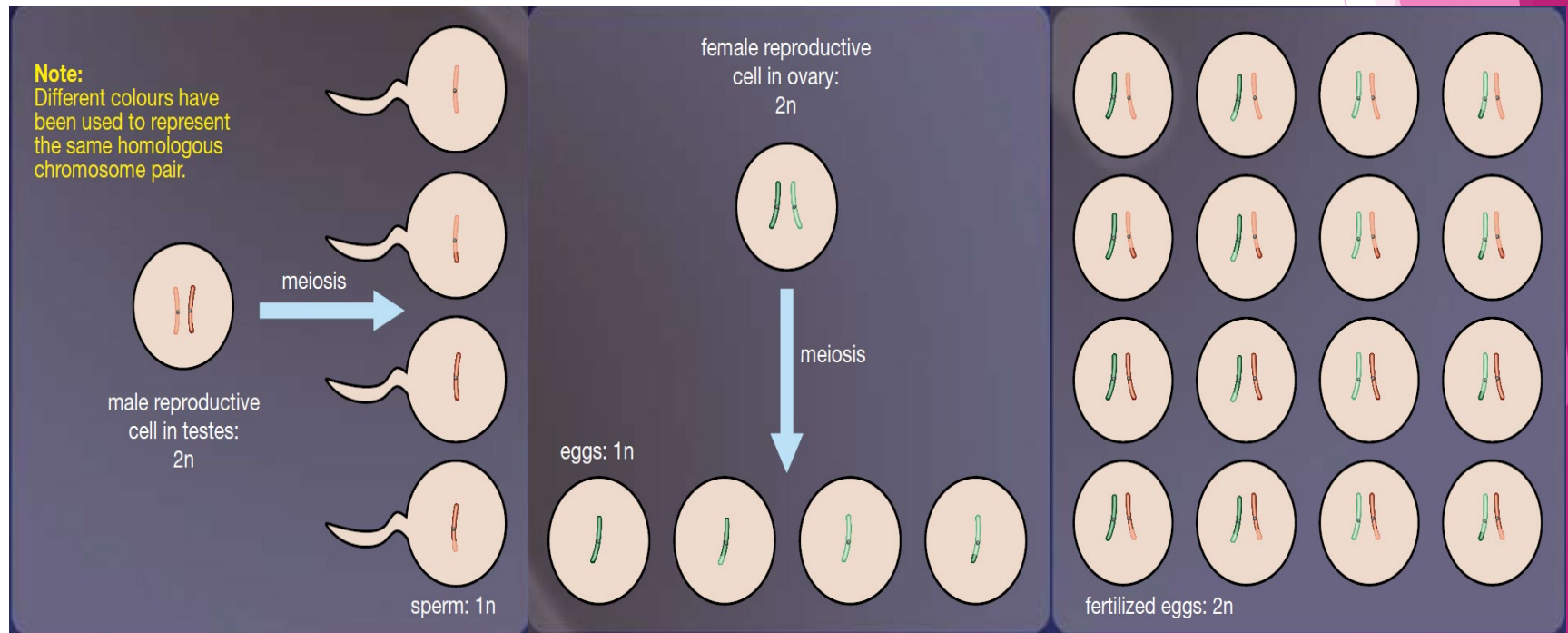


Meiosis vs. Mitosis

Characteristic	Mitosis	Meiosis
Original number of chromosomes per cell	23 pairs $2n = 46$	23 pairs $2n = 46$
Final number of chromosomes per cell	23 homologous pairs $2n = 46$	23 haploid cells $N = 23$
Number of cell divisions	1	2

Fertilization

- ▶ Once the male gamete meets the female gamete, **fertilization** happens and they create a zygote
- ▶ Each gamete brings half of the chromosomes needed for an autosomal cell, creating a genetically unique offspring
- ▶ Once two gametes meet they make a **zygote**



How many possibilities?

Number of Pairs of Chromosomes	Number of Possible Gametes Produced by One Parent (excluding crossing over)	Number of Possible Unique Combinations of Offspring Produced by Two Parents
n	2^n	$(2^n) \times (2^n)$
1	$2^1 = 2$	$(2^1) \times (2^1) = 2 \times 2 = 4$
2	$2^2 = 4$	$(2^2) \times (2^2) = 4 \times 4 = 16$
3	$2^3 = 8$	$(2^3) \times (2^3) = 8 \times 8 = 64$