Unit 3

Genetics

This unit online, with its interactives:
science-playground.com/s/9a3

You began as a single cell, smaller than the dot on this i. That cell held a set of instructions from your mother and another from your father, and every cell of your body has carried a copy of them ever since. The instructions are written in DNA, and the DNA is packed into **chromosomes**: 46 of them in nearly every cell of your body.

Stretched out end to end, the DNA of the 46 chromosomes in one of your cells would be about 2 meters long. It fits inside a nucleus only a few thousandths of a millimeter across, wound and folded so neatly that the cell can still find and read any part of it. This unit is about those chromosomes, the kinds they come in, and how they carry the **genes** that pass your characters on from one generation to the next. That study is called **genetics**.

3.1 Chromosome and its types

Look at the nucleus of a cell that isn't dividing, even under a good microscope, and you see only a fine, tangled network of threads. These threads are **chromatin**: long molecules of **DNA** wound around proteins called **histones**, like thread wound on many tiny spools. When the cell gets ready to divide, the chromatin coils and folds up tighter and tighter, until it forms short, thick bodies that can be seen clearly and counted. These are the **chromosomes**.

The name was given in 1888 by the German anatomist Wilhelm Waldeyer, from the Greek *chroma*, color, and *soma*, body: "colored bodies", because they take up stains so strongly that they stand out darkly in a stained cell.

A **chromosome** is a thread-like body in the nucleus, made of DNA and histone proteins, that carries the genes. Each chromosome is one single, very long DNA molecule. Before a cell divides, it copies every chromosome, so a chromosome seen at the start of cell division is double: two identical copies lying side by side, joined at one point.

Bacteria are different. A bacterium (a prokaryote) has no nucleus: its DNA is usually a single ring lying in the cytoplasm, not wound on histones as ours is. In this unit, a

DID YOU KNOW?

Packing it in

Human chromosome 1, the largest, is one DNA molecule of about 248 million base pairs. Each base pair adds 0.34 nm to the length, so stretched out it would be $248{,}000{,}000 \times 0.34\ \text{nm} \approx 8.4\ \text{cm}$ long. At cell division, it is coiled into a body only about a hundredth of a millimeter long, several thousand times shorter.

chromosome means the chromosome of a eukaryote, a plant, animal or fungus.

Structure of a chromosome

A chromosome is easiest to study at **metaphase** of cell division, when it is most tightly coiled and lines up in the middle of the cell. Then it looks like this:

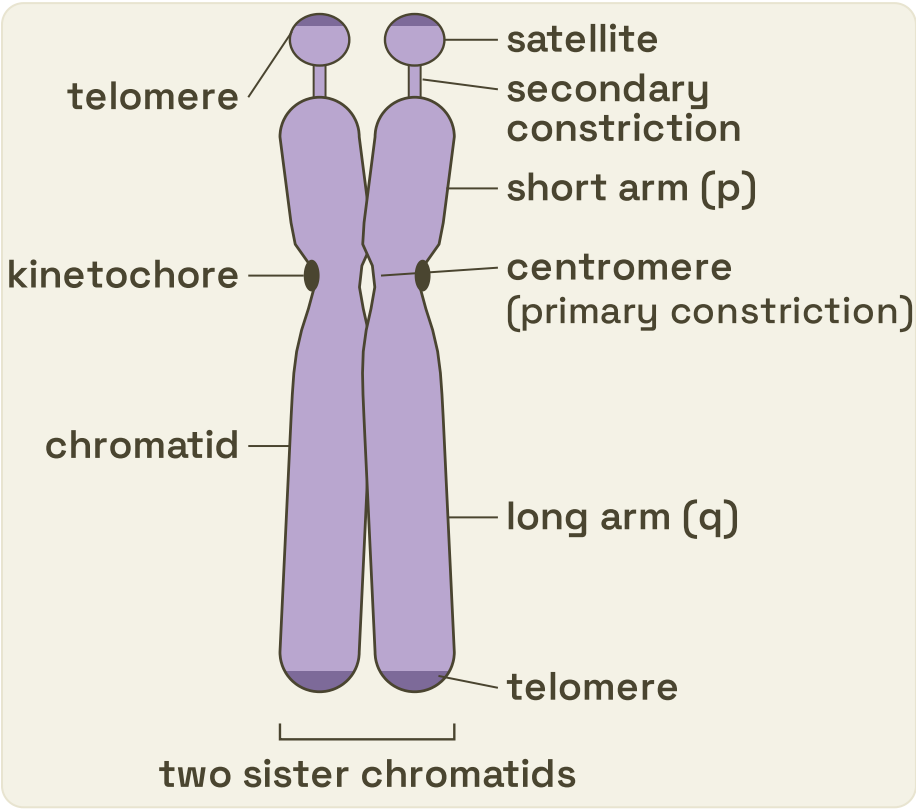


Figure 3.1 *The parts of a chromosome at metaphase. This one carries a satellite on its short arm; most chromosomes have none.*

1. Chromatids. A chromosome at metaphase is made of two identical halves called **chromatids**. They are copies of each other, made when the DNA was copied, so they are called **sister chromatids**. When the cell divides, they are pulled apart, and each one becomes a chromosome of one of the two new cells.

2. Centromere. The sister chromatids are held together at a narrow point called the **centromere**. It is also called the **primary constriction**, because the chromosome is pinched in there. On each side of the centromere is a small disc of protein, the **kinetochore**, where the spindle fibers attach during cell division. The fibers pull on the kinetochores, so the centromere is the part of a chromosome that leads the way to the pole.

3. Arms. The centromere divides each chromatid into two **arms**. Unless the centromere is exactly in the middle, one arm is shorter than the other. The short arm is called **p** (said to be for *petit*, French for small) and the long arm **q**, the next letter.

4. Telomeres. The two ends of a chromosome are its **telomeres**. In humans, a telomere is the same six bases, TTAGGG, repeated hundreds to thousands of times. Telomeres protect the ends of the chromosome, as the plastic tips of a shoelace keep it from fraying, and stop the ends of different chromosomes from sticking to each other.

5. Secondary constriction and satellite. A few chromosomes have a second narrow point besides the centromere: a **secondary constriction**. Many secondary constrictions hold the genes for making ribosomes, and the nucleolus forms around them, so they are also called **nucleolar organizers**. The small piece of chromosome beyond a secondary constriction, at the end of an arm, looks like a knob on a stalk and is called a **satellite**. A chromosome with a satellite is a **SAT chromosome**. In humans, chromosomes 13, 14, 15, 21 and 22 carry satellites on their short arms.

CAREFUL

Old names for the parts

Some books describe a chromosome as coiled threads (**chromonemata**) lying in a jelly (**matrix**) inside a skin (**pellicle**). That picture comes from the light microscopes of a century ago. The electron microscope showed no skin around a chromosome: a chromosome is its chromatin, DNA wound on histones, folded in loops onto a framework of other proteins. *Chromonema* is still sometimes used for the coiled chromatin thread. If an exam asks for the parts of a chromosome, name the chromatids, centromere, kinetochore, arms, telomeres, secondary constriction and satellite.

Check 1

At which part of a chromosome do the spindle fibers attach during cell division?

a the telomere
b the satellite
c the kinetochore, on the centromere
d the secondary constriction

Types of chromosome on the basis of structure

Chromosomes differ in size and shape, and the main way they differ in shape is **where the centromere is**. Its place decides how long the two arms are, and so how the chromosome looks when the spindle fibers drag it by its centromere at anaphase, its arms trailing behind. On this basis there are four types:

a. Metacentric chromosome. The centromere is in the middle, so the two arms are equal or nearly equal. At anaphase it is **V-shaped**. Examples: human chromosomes 1, 3, 16, 19 and 20.

b. Submetacentric chromosome. The centromere is a little to one side of the middle, so one arm is clearly longer, up to about three times the other. At anaphase it is **L-shaped**. Examples: human chromosomes 2, 4 to 12, 17, 18 and the X chromosome.

c. Acrocentric chromosome. The centromere is near one end, so the short arm is very short. At anaphase it is **J-shaped**. Examples: human chromosomes 13, 14, 15, 21, 22 and the Y chromosome.

d. Telocentric chromosome. The centromere is at the very end, so there is only one arm. At anaphase it is **rod-shaped**, like the letter I. Humans have none, but almost all of a house mouse's 40 chromosomes have their centromere at, or almost at, one end.

The type is set by the **arm ratio**, the length of the long arm divided by the length of the short arm:

$$\text{arm ratio} = \frac{\text{length of long arm } (q)}{\text{length of short arm } (p)}$$

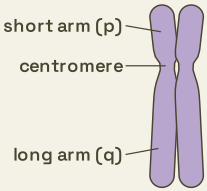
Type	Centromere	Arm ratio, q ÷ p	Shape at anaphase
Metacentric	in the middle	1 to 1.7	V
Submetacentric	a little off the middle	1.7 to 3	L
Acrocentric	near one end	more than 3	J
Telocentric	at the end	no short arm	I (rod)



metacentric
in the middle
p : q = 1 : 1



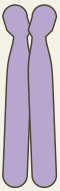
V-shaped



submetacentric
a little off the middle
p : q = 1 : 2



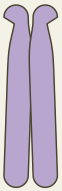
L-shaped



acrocentric
near one end
p : q = 1 : 5



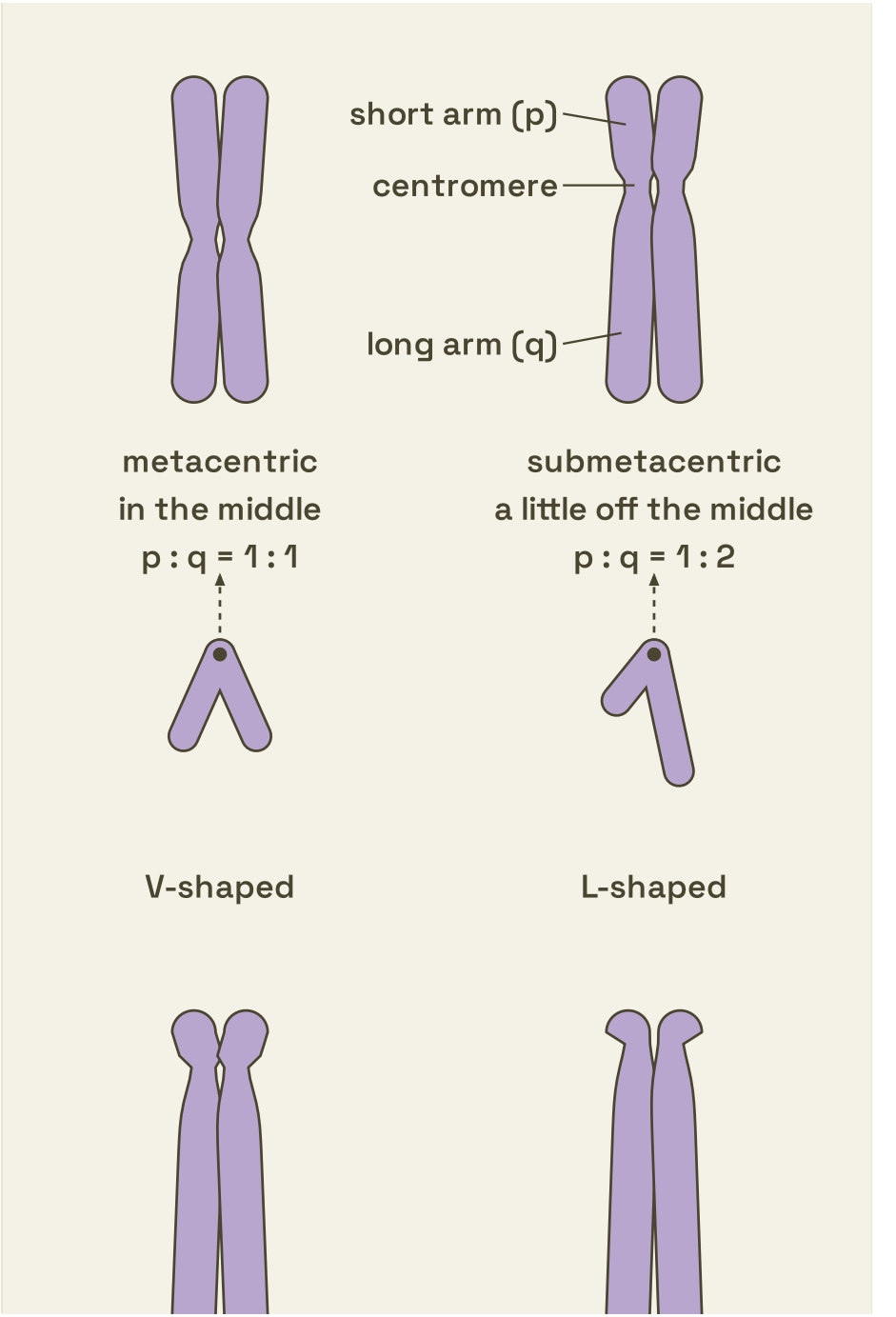
J-shaped



telocentric
at the very end
no short arm to speak of



I-shaped


short arm (p)
centromere
long arm (q)
metacentric
in the middle
p : q = 1 : 1
submetacentric
a little off the middle
p : q = 1 : 2
V-shaped
L-shaped

Some books also sort chromosomes by the **number of centromeres**: most have one (**monocentric**); a broken and wrongly joined chromosome may have two (**dicentric**) or none (**acentric**) and is torn or lost when the cell divides; and in a few living things, such as the worm *Caenorhabditis elegans*, the centromere runs along the whole length (**holocentric**).

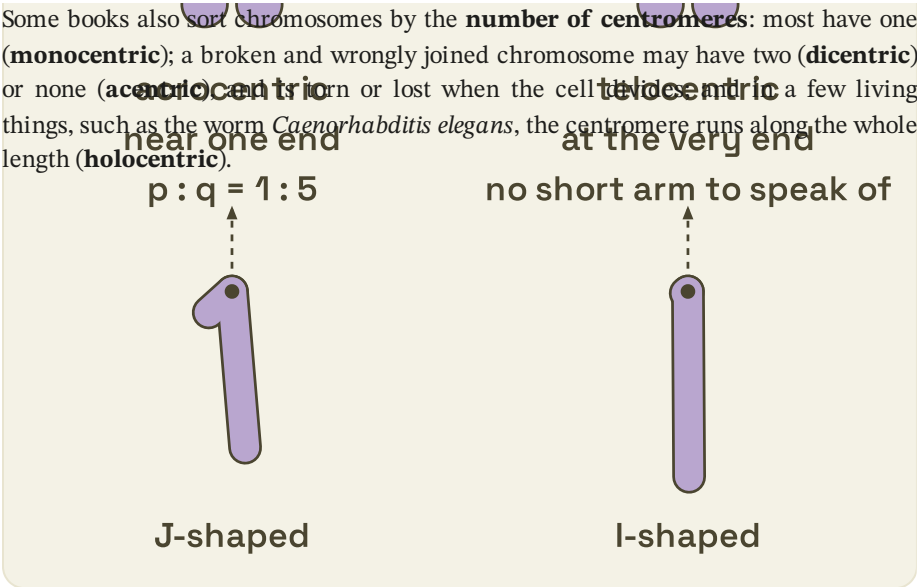


Figure 3.2 *The four types of chromosome, all drawn the same length, with where the centromere is under each name, and the short arm (p) and long arm (q) named on the second. Under each is one chromatid at anaphase, pulled up toward the pole by its centromere (the dot), its arms trailing behind: the place of the centromere decides its shape.*

CAREFUL

Where one type ends and the next begins

School books and exams use the four types as this unit does: **telocentric** only when the centromere is at the very tip, and **acrocentric** for every chromosome with an arm ratio over 3 whose centromere is not at the tip. Cytologists, who sort chromosomes for a living, use a finer scheme set out by Albert Levan and two colleagues in 1964. It keeps the boundaries of 1.7 and 3, then splits the near-the-end chromosomes in two: **subtelocentric** for an arm ratio of 3 to 7, and **acrocentric** only beyond 7, with telocentric for a centromere exactly at the end. So the same chromosome can have two names: human chromosomes 13, 14, 15, 21, 22 and the Y are acrocentric in the school scheme (and in the human list agreed at Denver in 1960), but subtelocentric in Levan's. In an exam, use the four school types.

WORKED EXAMPLE

Which type is it?

A chromosome at metaphase is 6.0 μm long, and its centromere is 1.2 μm from one end. Find its arm ratio and name its type.

Answer

Short arm, $p = 1.2$ μm. Long arm, $q = 6.0 - 1.2 = 4.8$ μm.

$$\text{arm ratio} = \frac{q}{p} = \frac{4.8}{1.2} = 4.0$$

An arm ratio of more than 3 means the centromere is near one end: **an acrocentric chromosome**, J-shaped at anaphase.

WORKED EXAMPLE

Finding the arms

A submetacentric chromosome is 9 μm long and its arm ratio is 2. How long is each arm?

Answer

Let the short arm be p. Then the long arm is $2p$, and together they make the whole length:

$$p + 2p = 9 \text{ μm} \quad \text{so} \quad 3p = 9 \text{ μm}, \quad p = 3 \text{ μm}$$

The short arm is 3 μm and the long arm 6 μm. Check: $6 \div 3 = 2$, and an arm ratio of 2 is between 1.7 and 3, submetacentric, as the question says.

YOUR TURN

A chromosome 8 μm long has its centromere 3.5 μm from one end. What type is it?

Answer

$p = 3.5$ μm, $q = 8 - 3.5 = 4.5$ μm, arm ratio $= 4.5 \div 3.5 \approx 1.3$, which is less than 1.7: **metacentric**.

DID YOU KNOW?

Your chromosome 2 was once two

Chimpanzees, gorillas and orangutans have 48 chromosomes; people have 46. Our large chromosome 2 matches two of the apes' smaller chromosomes joined end to end. The join can still be read in our DNA: in the middle of chromosome 2 there are telomere repeats, TTAGGG, facing each other, where two ends met, and the remains of a second, unused centromere.

Check 2

At anaphase, a chromatid is pulled toward the pole and makes a J shape. Its chromosome is:

a metacentric
b submetacentric
c acrocentric
d telocentric

Types of chromosome on the basis of function

By what they do, chromosomes are of two types.

a. Autosomes. These carry the genes for the characters of the body (height, skin color, blood group and thousands of others), and have little to do with whether an individual is male or female. The two chromosomes of each pair are alike in size, shape and the genes they carry. Of our 46 chromosomes, 44 (22 pairs) are autosomes. They are also called **somatic chromosomes**.

b. Sex chromosomes. These decide the sex of an individual. In humans there is one pair: **XX** in a female and **XY** in a male. The X is a large submetacentric chromosome with about 800 genes for proteins; the Y is a small acrocentric one with only a few dozen. Because the two members of the pair can be different, sex chromosomes are also called **allosomes** or **heterosomes** ("different bodies").

So a woman's body cell has **44 + XX** and a man's **44 + XY**. A picture of all the chromosomes of one cell, arranged in pairs by size, is called a **karyotype**.

Autosomes	Sex chromosomes
Carry genes for the body's characters.	Decide the sex of the individual.
22 pairs (44) in a human body cell.	1 pair (2) in a human body cell.
The two of a pair are alike in size and shape.	The two may differ (X and Y in a man).
The same in males and females.	XX in a human female, XY in a male.

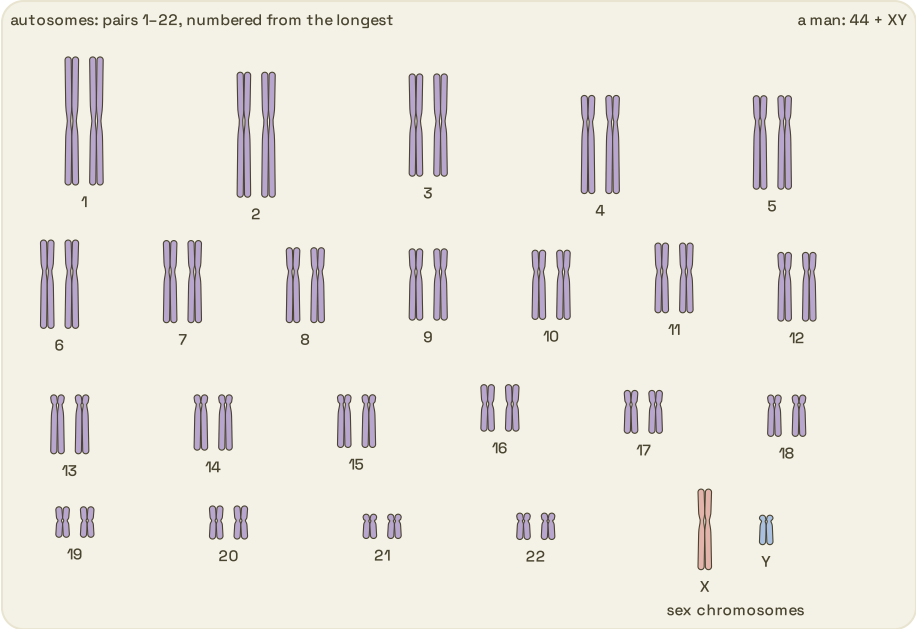


Figure 3.3 *A man's 46 chromosomes drawn as a karyotype: 22 pairs of autosomes, numbered from the longest, and the sex chromosomes X and Y. A woman's karyotype is the same, except that she has two X chromosomes.*

Not every living thing decides sex the same way. In birds, it is the female who has two different sex chromosomes, called Z and W (ZW), and the male who has two alike (ZZ). Most flowering plants, such as rice and the pea, have flowers that are both male and female, and have no sex chromosomes at all.



Figure 3.4 *A real karyotype of a man, photographed through a microscope. The stain gives each chromosome its own pattern of bands, which helps to match the pairs.*
Photo: Courtesy: National Human Genome Research Institute, Public domain

Every member of a species has the same number of chromosomes in its body cells, and it is usually even, because they come in pairs: one of each pair from the mother and one from the father. A body cell with two sets is **diploid** (2n); an egg or sperm, with one set, is **haploid** (n). A human body cell has 2n = 46, and a human sperm or egg n = 23: 22 autosomes and one sex chromosome.

A STORY FROM SCIENCE

Counting to 46

For more than thirty years, every biology book said that humans have 48 chromosomes, the number the American Theophilus Painter had counted in 1923. Human chromosomes are small and crowded, and they lay in a tangle that was hard to count. In December 1955, in Lund in Sweden, Joe Hin Tjio and Albert Levan used newer methods: colchicine to stop dividing cells at metaphase, and a weak salt solution that made the cells swell and spread their chromosomes apart. Tjio counted 46, again and again, in cell after cell. They published it in 1956. The correct number had been hiding in plain sight, and it took a better method, not better eyes, to see it.

Check 3

How many autosomes are there in a human egg cell?

a 44 **b** 22
c 23 **d** 46

3.2 Introduction to genetics

A calf looks like a cow, not a goat, and a child often has her mother's eyes or her grandfather's laugh. Characters pass from parents to their young: this is **heredity**. Yet no two children of the same parents are exactly alike, except identical twins, and even they differ a little. These differences between members of the same species are called **variation**.

Genetics is the branch of biology that studies heredity and variation: how characters pass from parents to offspring, and why offspring differ from their parents and from each other. The English biologist William Bateson coined the word, first in a letter in 1905 and then in public in 1906, from a Greek word meaning to give birth.

Some variation is **hereditary**: it is in the genes and can be passed on, like blood group or the shape of the earlobe. Some is **environmental**: it comes from how an individual lives, and isn't passed on. A wrestler's strong arms, a farmer's tanned face and a scar are not inherited by their children. Most characters, such as height, depend on both: the genes set the range, and food and health decide where in it a person ends up.

Mendel, the father of genetics

The rules of heredity were first worked out by Gregor Johann Mendel, a monk in the town of Brno (in today's Czech Republic), who crossed thousands of pea plants in his monastery garden from 1856 to 1863. He showed that characters are passed on by particles that come in pairs, one from each parent, and that they don't blend but stay separate, even when one is hidden for a generation. He published his results in 1866, but nobody understood how important they were. In 1900, sixteen years after his death, three botanists found the same rules on their own and then found his paper. Mendel is called the **father of genetics**.

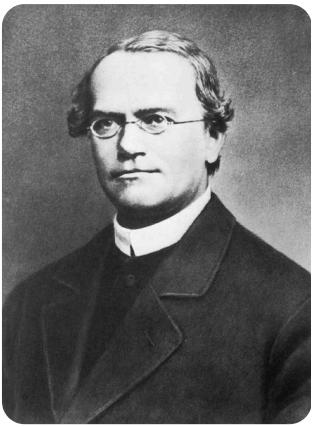


Figure 3.5 *Gregor Johann Mendel (1822–1884). He never saw a chromosome, yet his rules describe exactly how chromosomes are shared out to eggs and sperm.*
Photo: Unknown author, Public domain

The words you will need in genetics:

Gene: a part of a chromosome (a stretch of its DNA) that carries the instructions for one character, or more exactly for making one protein or one RNA. The Danish botanist Wilhelm Johannsen named it in 1909.

Allele: one of the different forms of the same gene. The gene for a pea plant's height has two alleles: one for tall (written T) and one for dwarf (t).

Dominant and recessive: when both alleles are present, the one that shows is **dominant** (T), and the one hidden is **recessive** (t).

Homozygous and heterozygous: an individual with two alike alleles (TT or tt) is **homozygous**, or pure; one with two different alleles (Tt) is **heterozygous**, or hybrid.

Genotype and phenotype: the **genotype** is the set of alleles an individual has (TT, Tt or tt); the **phenotype** is the character we see (tall or dwarf). TT and Tt plants have different genotypes but the same phenotype: both are tall.

Why genetics matters

Farming. Plant breeders cross varieties to bring together the alleles they want, such as high yield, resistance to disease or drought, or a short stem that doesn't fall over in the wind. The new rice, maize and wheat varieties grown in Nepal are made this way, and so are cows and buffaloes that give more milk.

Medicine. Some diseases, such as thalassemia, hemophilia and sickle-cell anemia, are inherited. Knowing how they are passed on lets doctors test for them and advise

families.

Identifying people. Everyone's DNA is different, except identical twins', so DNA can show who a child's parents are, or who was at the scene of a crime (DNA fingerprinting, in Grade 10 Optional Science).

Understanding life. Genetics explains why species vary, how they change over time, and how all living things are related.

Check 4

A child of two tall parents grows up shorter than both because he was badly fed as a small child. This difference is:

a hereditary variation, because it came from his parents

b environmental variation, which won't be passed to his children

c a mutation in his genes

d dominance of a dwarf allele

3.3 Relation between gene and chromosome

Mendel said characters are passed on by particles in pairs, but he had no idea where they were. Chromosomes were seen in the 1880s, but nobody knew what they did. The two discoveries came together in the early 1900s, and the link between them is the base of modern genetics: **genes are parts of chromosomes**.

A **gene** is a segment of the DNA in a chromosome. One chromosome is one very long DNA molecule, and it carries hundreds or thousands of genes, one after another along its length, like beads on a string. Each gene has its own fixed place on its own chromosome, called its **locus** (plural *loci*).

The chromosomes of a body cell come in pairs, and the two of a pair, called **homologous chromosomes**, carry the same genes at the same loci, one from the mother and one from the father. So every gene on an autosome is present twice in a body cell, once on each homologous chromosome. The two copies may be the same allele (TT) or different alleles (Tt). This is why genes come in pairs, as Mendel found.

The chromosome theory of inheritance

In 1902 and 1903, the American student Walter Sutton, watching chromosomes in grasshopper cells, and the German biologist Theodor Boveri, working with sea urchin eggs, saw that chromosomes behave exactly as Mendel's particles should:

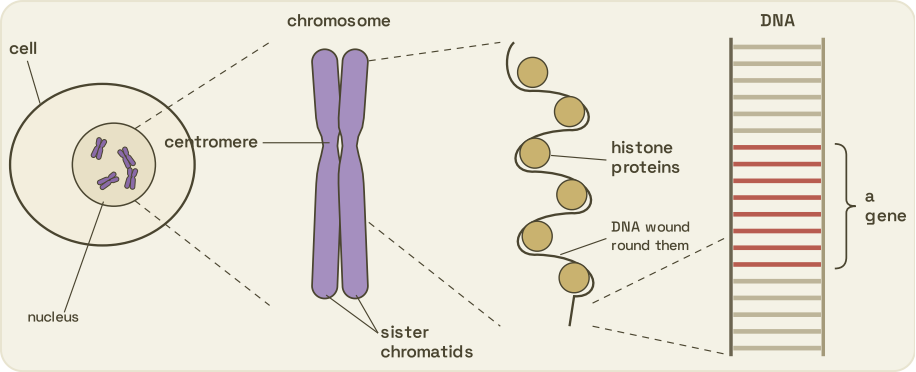


Figure 3.6 *From a cell to a gene. The nucleus holds the chromosomes; a copied chromosome is two sister chromatids joined at the centromere; each chromatid is one very long DNA molecule wound on histone proteins; a gene is one stretch of that DNA.*

Mendel's factors (genes)	Chromosomes
Come in pairs in a body cell.	Come in pairs in a body cell.
The two of a pair separate when gametes form, so each gamete gets only one.	The two of a pair separate in meiosis, so each gamete gets only one.
Each pair is shared out independently of other pairs.	Each pair lines up and separates independently of other pairs.
One of each pair comes from each parent.	One of each pair comes from each parent.

So, they said, the genes must be **on** the chromosomes. This idea is the **chromosome theory of inheritance**: genes are carried on chromosomes, and they pass from parents to offspring because the chromosomes do.

Morgan's students found that genes on the same chromosome tend to be inherited together (**linkage**), and one of them, Alfred Sturtevant, while still an undergraduate, used how often linked genes were separated to draw the first map of genes along a chromosome (published in 1913): they lie in a line, each in its own place.

A STORY FROM SCIENCE

Morgan's white-eyed fly

Thomas Hunt Morgan, in New York, doubted the chromosome theory, so he set out to test it on the fruit fly, *Drosophila*, which goes from egg to adult in about ten days and has only four pairs of chromosomes. In 1910, among his red-eyed flies, he found one male with white eyes. When he bred it, the white eyes turned up almost only in males, and passed from grandfather to grandson through the daughters, just as the X chromosome is passed on. The gene for eye color had to be on the X chromosome. It was the first gene ever placed on a particular chromosome, and Morgan, convinced at last, won the Nobel Prize in Physiology or Medicine in 1933 for showing the part chromosomes play in heredity.



Figure 3.7 *The fruit fly Drosophila melanogaster, about 3 mm long, which you may have seen around ripe bananas. Its red eyes are the usual kind; Morgan's mutant had white ones.*
Photo: André Karwath aka Aka, CC BY-SA 2.5

Gene and chromosome compared

CAREFUL

How many genes?

A human has about 20,000 genes that code for proteins, shared among 23 kinds of chromosome: chromosome 1 carries about 2,000, the small Y only a few dozen. Genes for proteins make up less than 2% of our DNA; much of the rest controls when genes are switched on, makes RNAs that aren't proteins, or has no known job. So "a chromosome is made of genes" is too simple: a chromosome **carries** genes, with a lot of other DNA between them.

Gene	Chromosome
A segment of DNA.	A whole DNA molecule with its proteins.
Carries the code for one protein or RNA, and so for a character.	Carries hundreds or thousands of genes.
Too small to be seen under a light microscope.	Seen under a light microscope at cell division.
Lies at a fixed place (locus) on a chromosome.	Lies in the nucleus.
About 20,000 kinds (for proteins) in humans.	23 kinds (46 in all) in a human body cell.

DID YOU KNOW?

Genes outside the chromosomes of the nucleus

Not quite every gene is in the nucleus. Each mitochondrion has its own tiny ring of DNA with 37 genes, and in plants each chloroplast has its own as well. You get your mitochondria from your mother's egg (a sperm's are destroyed), so your mitochondrial genes came from your mother, her mother, and her mother before her.

Check 5

Homologous chromosomes carry:

a the same alleles of every gene

b the same genes at the same loci, though the alleles may differ

c different genes, one set from each parent

d only the genes for sex

IN SHORT

- A **chromosome** is a thread-like body in the nucleus, one long DNA molecule wound on histone proteins, carrying genes. It is seen clearly at cell division.
- At metaphase it has two **sister chromatids** joined at the **centromere** (primary constriction), with **kinetochores** where spindle fibers attach, a short arm **p** and a long arm **q**, **telomeres** at the ends, and on some chromosomes a **secondary constriction** and **satellite**.
- **By structure** (the centromere's place, measured by the arm ratio q ÷ p): **metacentric** (middle, V), **submetacentric** (off the middle, L), **acrocentric** (near the end, J), **telocentric** (at the end, I).
- **By function: autosomes** (22 pairs in humans) carry genes for the body's characters; **sex chromosomes** (1 pair: XX female, XY male) decide sex.
- **Genetics** is the study of heredity and variation; Mendel is its father.
- A **gene** is a segment of DNA at a fixed **locus** on a chromosome; a chromosome carries many genes in a line. Homologous chromosomes carry the same genes at the same loci, so genes come in pairs.
- **Chromosome theory of inheritance:** genes are carried on chromosomes, which explains Mendel's rules; Morgan's white-eyed fly placed a gene on the X chromosome.

Exercise

1. Choose the correct option

a. The part of a chromosome that holds the two sister chromatids together is the:

a telomere
b centromere
c satellite
d chromonema

b. Which type of chromosome is not found in a normal human cell?

a metacentric
b submetacentric
c acrocentric
d telocentric

c. The term "genetics" was coined by:

a Gregor Mendel
b William Bateson
c Thomas Hunt Morgan
d Wilhelm Johannsen

d. The fixed place of a gene on a chromosome is called its:

- a allele
- b locus
- c kinetochore
- d centromere

e. The chromosome theory of inheritance was put forward by:

- a Mendel and Bateson
- b Sutton and Boveri
- c Watson and Crick
- d Tjio and Levan

2. Give reasons

a. Chromosomes are studied at the metaphase of cell division.

b. Chromosomes are found in pairs in a body cell.

3. Differentiate between

i. Metacentric and acrocentric chromosomes

ii. Heredity and variation

4. Answer the following questions

a. What is a chromosome? Draw a labeled diagram of a chromosome at metaphase.

b. Write the functions of the centromere, the telomeres and the secondary constriction.

c. Classify chromosomes on the basis of the position of the centromere. Draw each type.

d. Explain the relation between gene and chromosome.

e. State the chromosome theory of inheritance. Give any three points in which genes and chromosomes behave alike.

5. Work it out

a. A chromosome 7 μm long has an arm ratio of 2.5. Find the length of each arm and name the type of chromosome.

b. In a chromosome 12 μm long, the centromere is 5.5 μm from one end. Find the arm ratio and the type.

c. A dog's body cell has 78 chromosomes, and dogs decide sex by X and Y as humans do. How many autosomes are there in a dog's body cell, and how many chromosomes, and which, in its sperm?

d. A human cell is at metaphase of mitosis. How many chromosomes, chromatids and centromeres does it have?

Found a mistake? Please tell us.