Unit
2A

Chapter 20
Sources of variation in humans

Figure 20.1 Variation in humans

Unit content

Variation and evolution
New genetic combinations are made as a result of meiosis and fertilisation, giving rise to unexpected variations.

Variation from meiosis:

- crossing over
- random assortment
- non-disjunction.

Variation from fertilisation:

- random fertilisation.

What is variation?

Look at the people shown in Figure 20.1. They all belong to the same species, *Homo sapiens*; they are all humans. All have a similar body shape, two arms, two legs, a relatively flat face with two eyes, a nose, a mouth and so on. They have more in common with each other than with the members of any other species.

Despite all the basic similarities between people there are many differences. Can you see any two people in Figure 20.1 who are identical? Is there anyone in your class who is identical to you? Even identical twins have small differences between them; those who know them well can usually tell them apart.

Humans differ in a lot of ways, such as height, weight, eye colour, skin colour, body proportions. In addition to differences in visible characteristics humans differ in things like blood group, thought patterns, ability to taste and to smell, ability to solve problems, tolerance of heat and cold and resistance to disease.

All species show differences between individuals. The differences between members of a species are called **variation**. The people in Figure 20.1 show variation because they are all different.

Variation between one person and another is partly the result of environmental factors. Over your lifetime you will be exposed to a range of environmental factors, some of which will affect your appearance. For example, your basic skin colour was inherited from your parents but if you are light skinned, exposure to the sun or other environmental factors, may result in modification to this colour. If you have had an infectious disease, or have been vaccinated, you will have developed immunity to the disease. This is another example of an environmental influence changing your characteristics.

In this chapter we will not discuss the effects of the environment but will concentrate on the variation that occurs between inherited characteristics. What causes inherited variation? Why do you resemble your parents in some ways but are not identical to either of them? Variation in inherited characteristics comes about as a result of meiosis, random fertilisation and mutations.

Random assortment of chromosomes during meiosis

In Chapter 18 the principles of inheritance put forward by Mendel were discussed. Mendel found that the characteristics he was studying were inherited independently. That is, a plant with green pods could have either round or wrinkled seeds. Likewise, a plant with yellow pods could also produce either round or wrinkled seeds. Inheritance of pod colour had no effect on inheritance of seed shape and vice versa. Mendel decided that each individual must have two factors (genes) for a characteristic and that during the formation of the gametes (in humans, the eggs and the sperm), the pairs of factors separate. Each gamete receives only one set of factors, or genes, the other set going to another gamete. Mendel called this the principle of independent assortment.

When two gametes unite at fertilisation, the resulting cell has a combination of genes that is different from either parent. Gametes are formed by the type of cell division known as meiosis, which was described in Chapter 5. Each cell has two sets of chromosomes: one set from the male parent (the paternal chromosomes) and one from the female parent (the maternal chromosomes). Thus the chromosomes exist in **homologous** pairs—the genes on one member of the pair control the same characteristics as the genes on the other member of the pair. During the first meiotic division the homologous pairs of chromosomes separate. When these pairs of chromosomes separate, they do so at random. One member of each pair moves to

one pole of the cell, while the other member of the pair moves to the opposite pole. This results in 23 chromosomes moving to each pole of the cell.

An important feature of meiosis is that when the chromosomes move apart during the first meiotic division they do so independently. The way one pair of chromosomes separate is unaffected by the way any of the other pairs separate. For example, the copy of chromosome 1 that an egg cell receives in no way influences which of the two possible copies of chromosome 5 it gets. This random assortment takes place for each of the 23 pairs of human chromosomes. That means any single human egg receives one of two possible chromosomes 23 times. The total number of different possible chromosome combinations is 2^{23} which is approximately 8.4 million. And that's just for the eggs! The same random assortment goes on as each sperm cell is produced. Thus, when a sperm fertilises an egg, the resulting fertilised cell contains a combination of genes arranged in an order that has probably never occurred before and will probably never occur again.

Figure 20.2 shows a cell with just three pairs of chromosomes. Meiotic division of such a cell could produce 2^3 (i.e. 8) different types of gametes.

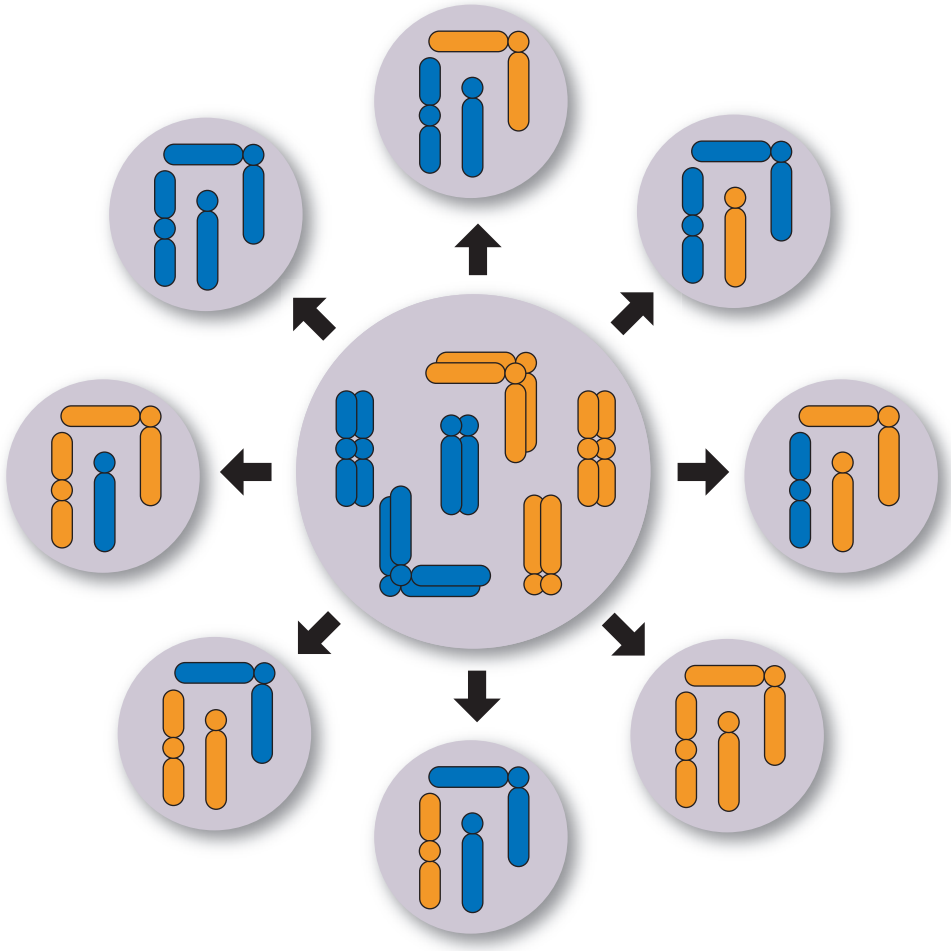

Figure 20.2 There are 8 possible combinations of chromosomes in gametes produced by a cell with just six chromosomes (three pairs)

Crossing over

Remember that when chromosomes become visible at meiosis they are already duplicated and each consists of a pair of chromatids joined at the centromere. When the homologous chromosomes pair during the first division of meiosis the chromatids may get tangled with one another, a situation known as **crossing over**

(see Fig. 20.3). The point where two chromatids cross is called a **chiasma** (plural, **chiasmata**). When crossing over occurs, the chromatids may break and reattach to a chromatid from a different chromosome. When this occurs, the result is a new combination of alleles along the chromosome (Fig. 20.3 and Table 20.1). This is called **recombination**.

To see a video of how crossing over takes place go to http://www.layyous.com/Videoclips/crossing_over.htm

If a gamete containing a chromosome with a recombined set of alleles is involved in fertilisation, the resulting offspring will have the new combination of alleles. This combination of alleles is not the same as that possessed by either of the parents.

Tracking the movement of alleles during crossing over helped scientists determine roughly how far apart two genes were on a chromosome. Since there are more chances for a break to occur between the alleles of two genes that lie far apart, it is more likely that one allele will stay on the original chromosome, while the other crosses over. Therefore, alleles that lie far apart are more likely to end up on two different chromosomes. On the other hand, alleles that lie very close together are far less likely to be separated by a break and to cross over. By examining the number of offspring with a recombination of alleles, scientists were laboriously able to map the order of genes on a chromosome. Today DNA analysis techniques are used to determine the genome.

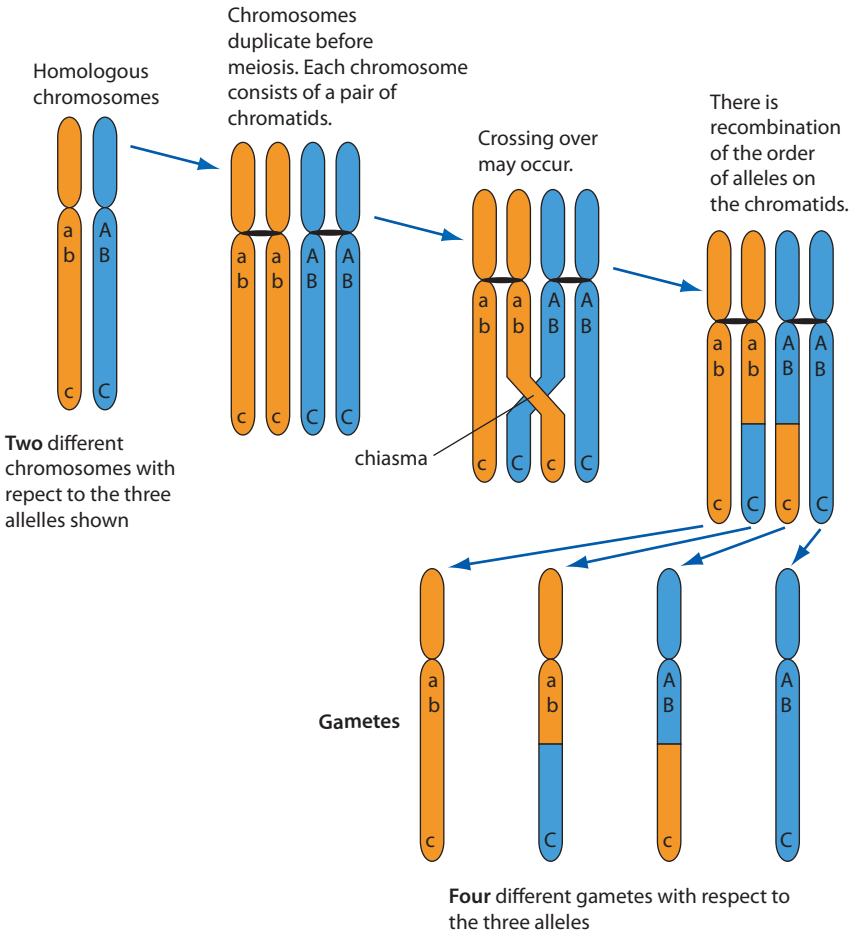

Figure 20.3 Crossing over and recombination during meiosis

Table 20.1 Crossing over changes the order of alleles on a chromatid

Order before crossing over	a b c	a b c	A B C	A B C
Order after crossing over	a b c	a b C	A B c	A B C

Non-disjunction

During the first division of meiosis, the homologous chromosomes pair and then separate. Sometimes one or more of the chromosome pairs may fail to separate when the cell divides. In the second meiotic division, one or more of the chromatids may fail to separate. These situations are called **non-disjunction**, and they result in one of the daughter cells receiving an extra chromosome and the other daughter cell lacking that chromosome (Fig. 20.4). In humans, if non-disjunction occurs in one of the chromosome pairs during meiosis, the resultant gametes would have either 24 chromosomes or 22 chromosomes, instead of the normal 23. After fertilisation with a normal gamete from the opposite sex, the zygote produced would have either 47 or 45 chromosomes, respectively. This would produce quite unexpected characteristics in the offspring. Usually, such changes to the chromosome number cause severe and distinctive birth defects and miscarriage often occurs early in the pregnancy.

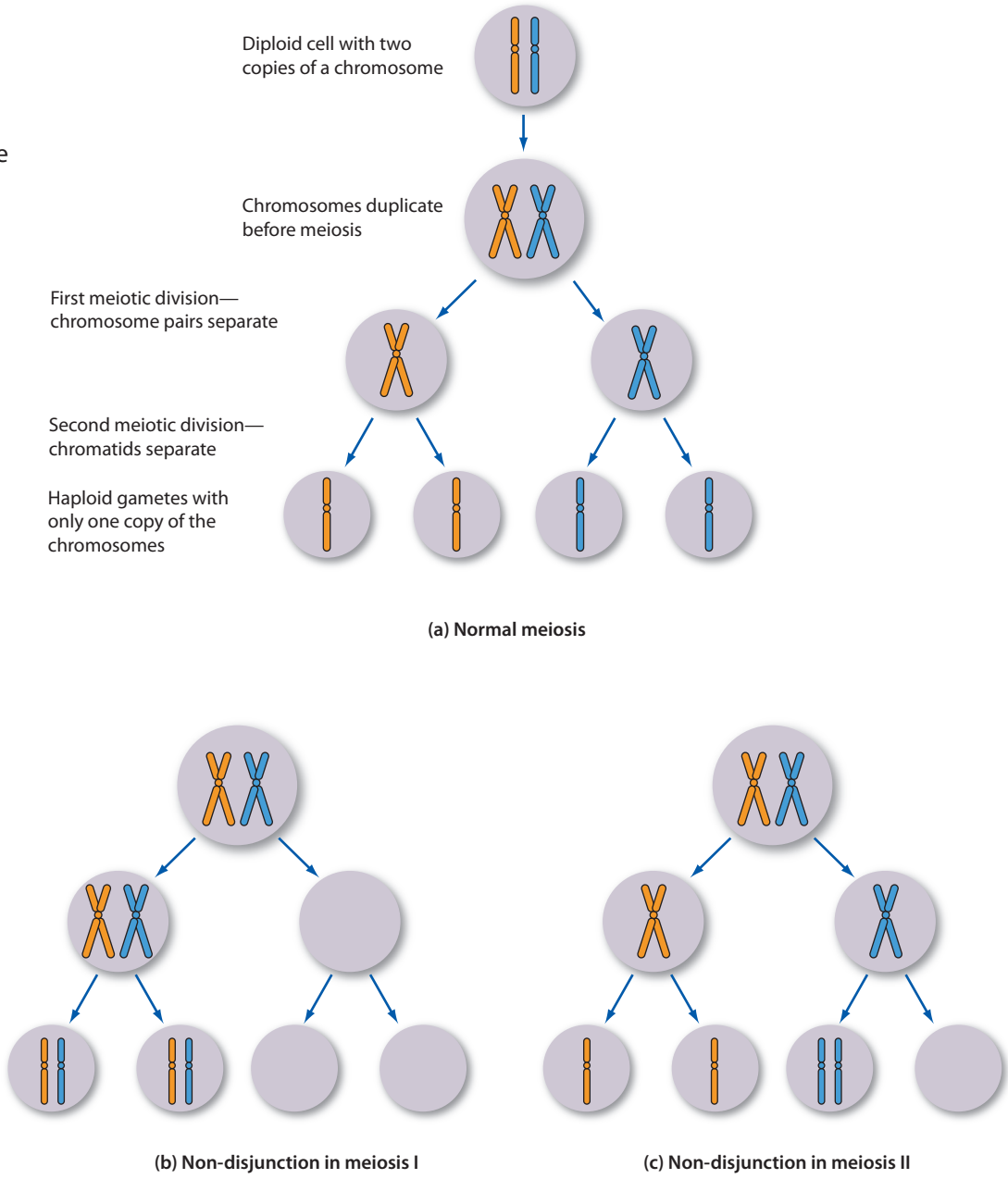

Figure 20.4
(a) Normal meiosis; **(b)** non-disjunction in the first meiotic division; **(c)** non-disjunction in the second meiotic division

Trisomy is where an individual inherits an extra copy of a chromosome—three copies instead of the normal two. One such chromosomal defect that occurs relatively frequently, especially in children of older mothers, is **Down syndrome**, or **trisomy 21** (Fig. 20.5). In this disorder an extra copy of chromosome 21 results in a characteristic facial appearance, variable degrees of mental retardation and physical abnormalities. Figure 20.6 shows the karyotype (the appearance—size, shape, and number —of the chromosomes in a cell) of this defect.

There are cases where trisomy occurs with other human chromosomes. An extra chromosome 13 produces individuals with mental retardation, a small head, an extra finger on each hand, a cleft palate and/or cleft lip, and malformations of the ears and eyes. An extra chromosome 18 results in the individual suffering from mental retardation and defects in the eyes, ears, hands and head.

Monosomy is where an individual is missing a chromosome—they have only one copy instead of the normal two. Like trisomy, monosomy usually results in severe malformations and often miscarriage.

Partial monosomy and partial trisomy can also occur. In **partial monosomy** part of a chromosome is missing—part of the chromosome has two copies, but part has only one copy. **Partial trisomy** occurs when part of an extra chromosome is attached to one of the other chromosomes. Partial trisomy 21 can result in many of the symptoms of Down syndrome.

Figure 20.5 A Down syndrome child with her mother

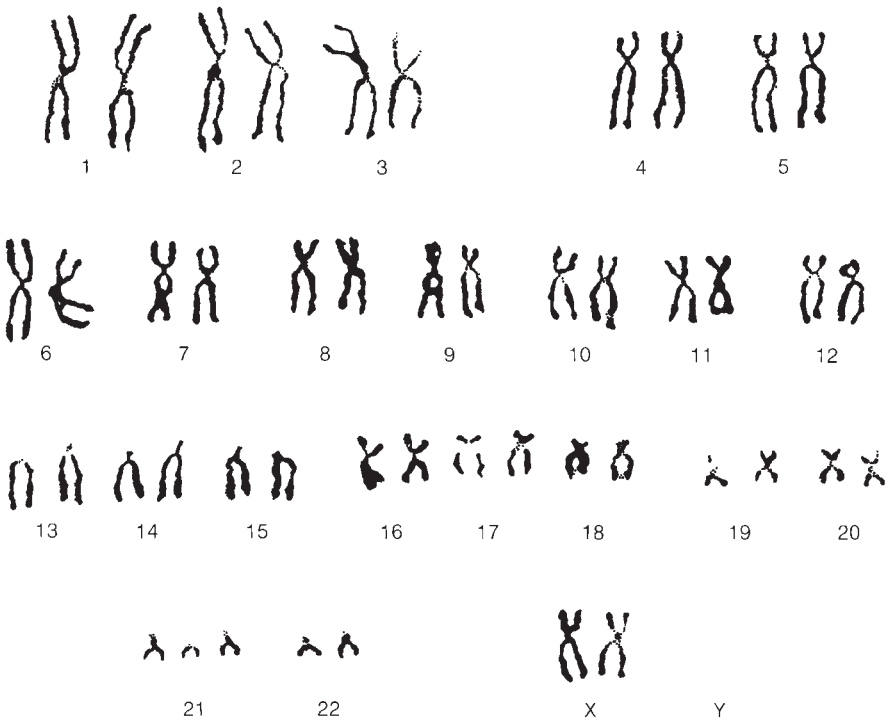

Figure 20.6 Karyotype of Down syndrome (note the extra chromosome 21)

EXTENSION

Turner's, Klinefelter's and Cri-du-chat syndromes are also chromosomal disorders.

Find out:

- about the causes and symptoms of each syndrome
- in what important ways these disorders differ from Down syndrome.

Random fertilisation

Earlier in this chapter the way pairs of homologous chromosomes move apart during the first meiotic division was discussed. That is, they separate independently of each other so that when the egg cells and sperm cells are produced, there is a huge range of combinations of the original maternal and paternal chromosomes.

Fertilisation occurs when a sperm joins with an egg to form the first cell of a new individual, the zygote. There is no way of determining exactly which sperm cell will fertilise any particular egg. **Random fertilisation**, in which any kind of sperm cell from the male parent can fuse with any kind of egg from the female parent, is therefore another source of variation that occurs as a result of sexual reproduction.

Females usually only release one egg at a time, but males produce hundreds of millions of sperm, each of which is slightly different in terms of the genetic material it carries. In addition, each sperm released into the female's reproductive tract has a different survival rate, so which one of the hundreds of millions of sperm will actually unite with the egg at the time of fertilisation depends completely on chance. Thus, more variability in the offspring is introduced depending on which sperm or egg is successful in fertilisation. The resulting offspring therefore has a completely different set of alleles from those of either of its parents.

Mutations

A mutation is a permanent change in the DNA that makes up a chromosome. Such a change leads to the sudden occurrence of new and different characteristics in offspring. Mutations will be discussed in detail in Chapter 21.

Accepting human variations

Humans vary in a host of characteristics as we have seen. Despite these variations, all humans are classified in the same species—*Homo sapiens*.

A **species** is a group of organisms that, having many characteristics in common, are able to interbreed and produce fertile offspring under natural conditions. The members of a species are reproductively isolated from other groups; that is, they cannot, or will not, mate with organisms from another species. Members of the human species are all basically alike, and thus able to interbreed and produce fertile offspring, but they are not capable of interbreeding with members of other species.

Because there is such variation within the human species, scientists have tried to classify humans into groups, or races, based on physical characteristics. Classification is an important scientific activity. Placing things in groups with common characteristics makes them easier to study and helps to show relationships between groups. Thus, in chemistry, we have the periodic table for the classification of the elements; in geology, rocks are classified according to their origins; in astronomy, pulsars and quasars are just two of the types of stars; and in biology, there is the classification of living things into kingdom, phylum, class, order, family, genus and species.

Attempts to classify the peoples of the world into races resulted in groupings with so many exceptions that the classification was unworkable. It is difficult to distinguish distinct groups of humans because migrations and intermarriages have blurred any genetic differences between populations. Such intermixing of people dates back to prehistoric times, but it is even more common today with modern transportation, which has increased human mobility. If one or two physical characteristics are selected, it may be possible to place people into reasonably distinct groups, but the greater the range of characteristics considered, the more impossible the task becomes. This

is because the amount of genetic diversity within populations is just as great as the diversity between them. Most biologists have therefore concluded that the classification of humans into races serves no useful purpose and is likely to increase conflict between human groups.

There are hundreds of different characteristics that occur within the human species, and so you can readily appreciate the huge number of combinations of characteristics possible in human beings. Differences in inherited characteristics are due to differences in the DNA in the nucleus of the cells. DNA profiling is now giving human biologists an insight into variation at the molecular level. Such studies show that humans are even more variable than previously thought. The genetic diversity is so great that every human who has ever lived is genetically unique.

What causes this variation? This chapter has examined how the random assortment of chromosomes during meiosis, together with random fertilisation of gametes, are major contributors to the enormous amount of variability seen in human populations.

Working scientifically

Activity 20.1 Modelling independent assortment

You will need (for each pair)

A large sheet of paper or laminated board or bench top to write on; 16 pipe cleaners (eight each of two different colours); wire ties; pencil or whiteboard marker; eraser; a coin

What to do

1. Read through Activity 5.2. The steps described will form the basis for this activity.
2. Draw a large outline on your paper to represent a cell and inside the cell draw a smaller circle to represent the nucleus.
3. The pipe cleaners will be your chromosomes. To model the chromosomes take four pipe cleaners of one colour to represent those from one parent (the maternal chromosomes) and four of another colour for chromosomes from the other parent (the paternal chromosomes).
4. Make each of the chromosomes that are the same colour different lengths so that you can distinguish them. For the colour you have decided came from the female parent, refer to the longest length as 'A', the next longest as 'B', and so on. For the colour you have decided came from the male parent, refer to the various lengths as 'a', 'b', etc.
5. Place a wire tie around each chromosome to represent the centromere. Before cell division the chromosomes duplicate so duplicate your chromosomes with a pipe cleaner of the same colour and length. Link the duplicates (chromatids) together using the wire tie (centromere). Label your duplicates with the same letter as the original. Place all your chromosomes in your outline of a cell. Your cell should now look like Figure 20.7.

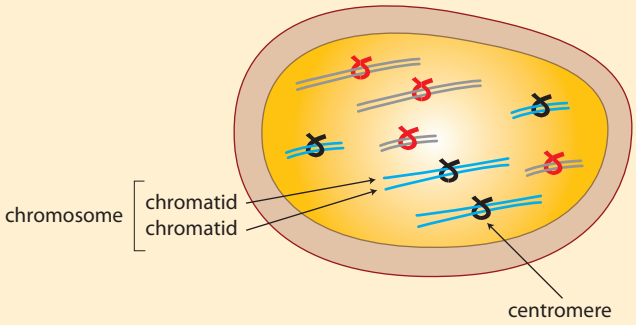

Figure 20.7 The model cell with eight duplicated chromosomes

6. At meiosis the like chromosomes pair off and arrange themselves across the centre of the cell attached to a spindle. Arrange your chromosomes in pairs by placing those of equal length together.
7. At the first meiotic division the members of the chromosome pairs separate. Toss your coin to decide to which end of the cell each of the chromosomes will migrate by deciding 'heads' for one end and 'tails' for the other. Be consistent for all your 'tosses'. Toss your coin four times to determine the direction of migration for the maternal chromosome; the paternal chromosome will move to the opposite pole.
8. Record the letters of the various chromosomes at each pole by completing rows 1 to 4 in a table similar to the one below.

	Chromosome combinations	
	'Head' end	'Tail' end
1		
2		
3		
4		
5		
6		
7		
8		

Independent assortment of chromosomes occurs at the first meiotic division so there is no need to simulate the second division.

9. Simulate another first meiotic division by repeating steps 6 to 8 and recording the results in rows 5 to 8 of your table.

Studying your data

1. Are all your combinations of chromosomes different? Are there any combinations that you think are missing?
2. How many combinations of these four chromosomes are possible? Remember that each cell receives one of two possible chromosomes four times (see page 270).
3. Combine your data with the other groups in the class to obtain a bigger sample. Are all the possible combinations for the four chromosomes now listed?

Interpreting your data

1. How has this activity indicated the variability brought about by independent assortment of the chromosomes?

2. If you got the same combination of chromosomes more than once, what does this imply about the first stage of meiosis?
3. Write a brief statement to summarise the variability brought about by the random assortment of chromosomes.

Activity 20.2 Independent assortment and fertilisation

Due to the independent assortment of chromosomes during meiotic division the number of different combinations of maternal and paternal chromosomes that are possible is equal to 2^n, where n is the haploid number of chromosomes. Thus, for the mosquito, which has 6 chromosomes the number of possible combinations of chromosomes in the gametes is $2^3 = 8$. At fertilisation, any two gametes may combine so the number of possible combinations of chromosomes in the offspring of mosquitoes is $8 \times 8 = 64$.

1. The fruit fly, *Drosophila*, has a chromosome number of 8. How many different combinations of chromosomes would be possible in the gametes of *Drosophila*?
2. How many different combinations of chromosomes would be possible in the offspring of *Drosophila*?
3. Kangaroos and the peas on which Mendel worked have 14 chromosomes. How many different combinations of chromosomes would be possible in the gametes of kangaroos and peas?
4. How many different combinations of chromosomes would be possible in the offspring of kangaroos and peas?
5. Beans and rock wallabies have a chromosome number of 22. Calculate the number of possible combinations of chromosomes in the gametes and the offspring of rock wallabies and beans.
6. *Homo sapiens* have a chromosome number of 46. The number of possible combinations of chromosomes in sperm or eggs is therefore 2^{23}, which is 8 388 608. The number of possible combinations of chromosomes in human offspring is 8 388 608 multiplied by 8 388 608. If crossing over is taken into account the total number of different ways that human gametes can combine is 2^{52}—a number that is greater than the total number of atoms in the solar system. Explain why no two humans (apart from identical twins) ever have exactly the same combination of alleles.

REVIEW QUESTIONS

1. What is meant by the term variation? Illustrate you answer with at least two examples.
2. Describe the events that take place during meiosis that lead to the random, or independent, assortment of chromosomes.
3. What is crossing over? Explain how it results in new combinations of characteristics in offspring. Draw a diagram to illustrate your answer.
4. Explain how non-disjunction takes place. Give an example of a common instance of this chromosomal defect.
5. Describe how random assortment contributes to variation in the offspring produced by sexual reproduction.

6. Explain how the joining of gametes at fertilisation contributes to variation.
7. Why do scientists no longer try to classify the human species into different races?

APPLY YOUR KNOWLEDGE

1. When a sperm fertilises an egg, the resulting fertilised cell contains a combination of genes arranged in an order that, in all probability, has never occurred before and is highly unlikely to occur again. What processes contribute to the uniqueness of the fertilised cell?
2. Look at the picture of human chromosomes (Fig. 18.5 on page 245). Which of the human chromosomes would be likely to have the greatest amount of crossing over? Explain the reason for your answer.
3. The relative location of genes along a chromosome affects the chances of them being separated by the process of crossing over. Use references to find out which human characteristics are more likely to be affected by crossing over.
4. The frequency of non-disjunction of chromosome 21 (Down syndrome) increases with the age of the mother. Find out how it is thought age contributes to non-disjunction.
5. In Australia, do you think that selection of a partner is a completely random event? Is there a range of social factors that may influence the choice of a partner and therefore contribute to the phenotypes found in the next generation?
6. Explain why independent assortment of chromosomes occurs at the first meiotic division and not the second.
7. Variation only occurs when organisms reproduce sexually. When a single-celled organism, like an *Amoeba*, reproduces asexually, the two new *Amoebae* are identical to the parent. Explain why asexual reproduction does not produce variation.
8. Mendel studied the inheritance of characteristics that just happened to be controlled by genes on different chromosomes. Explain why he would not have been able to arrive at the principle of independent assortment had he studied characteristics controlled by genes on the same chromosome.
9. Many people believe that the use of the term 'race' to describe variations in humans should not be permitted. In what ways could reference to race be harmful in our society?