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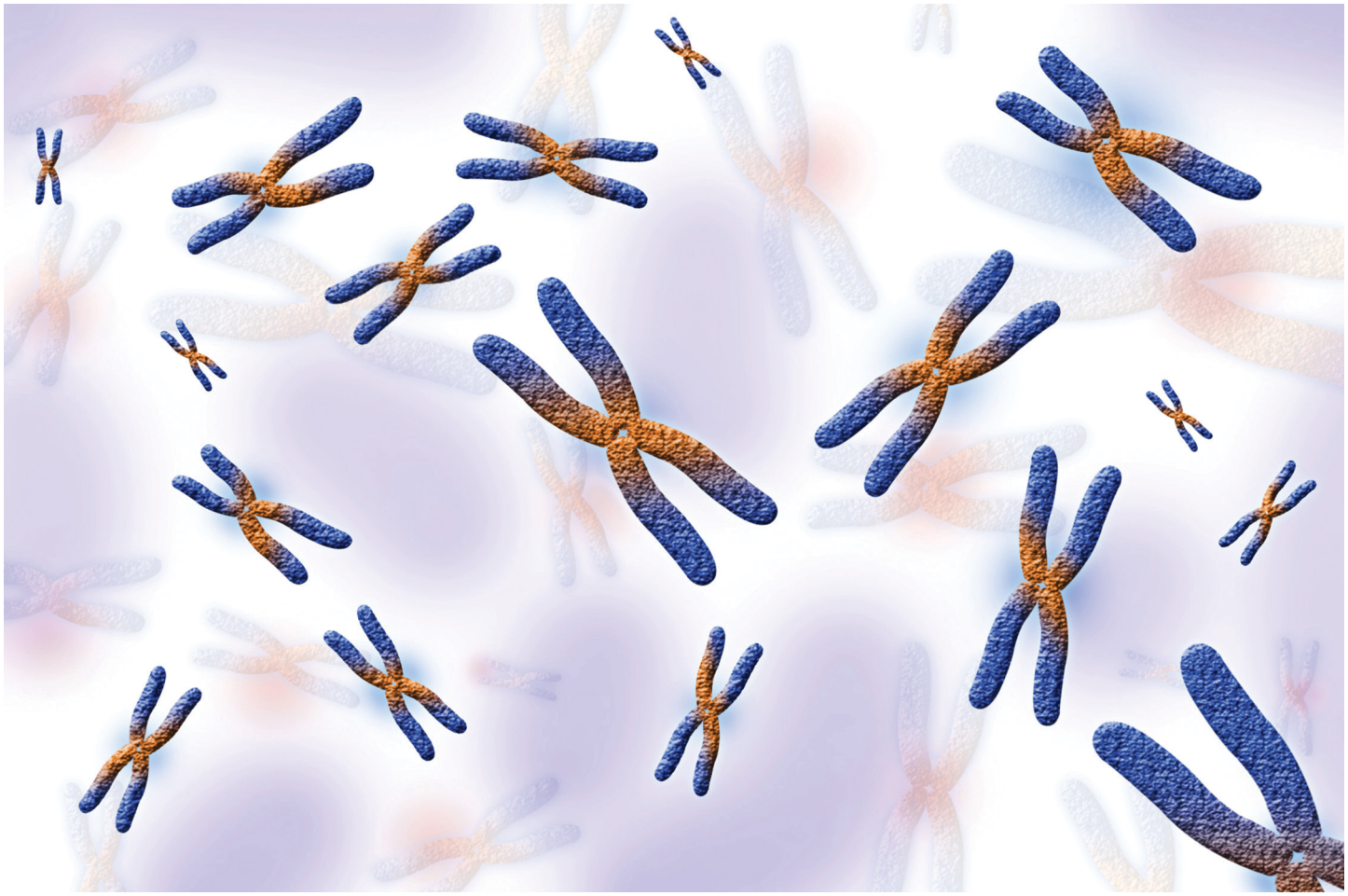
Inheritance of characters



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Contents

Introduction

Genes are units of inheritance that contribute to a person's behaviour and health. In this course you will learn what genes, DNA and chromosomes are and how they combine to make the human genome. You will also learn how the principles of inheritance work, the effect that our genetic make-up has on health, and how genetic material is passed on from generation to generation.

This OpenLearn course provides a sample of Level 1 study in Science.

Learning Outcomes

After studying this course, you should be able to:

- understand that genes are the units of inheritance for individual characteristics and also may contribute to susceptibility to certain diseases
- understand the number of chromosomes that make up the human genome and where they are located within the cell
- understand something of the immense scale of the human genome project
- understand how gametes are produced by the process of meiosis and how the full complement of 46 chromosomes is restored at fertilisation
- understand the role of the X and Y chromosomes in determining sex and how they are inherited.

1 Overview

1.1 Human genetics and health issues

1.1.1 Inheritance of characters

Imagine you have found some old family photograph albums which span many generations. What are the distinctive family features, or characters, that demonstrate the relatedness of individuals? In other words, what characters do they have in common? For example, they might have brown eyes, a white forelock in their hair, ears that are closely attached to the head, that is, without lobes. But you will also notice the striking differences between related individuals. For example, they may differ in height or hair texture. Some similarities and differences extend to characters that are not visible. For example, individuals may share the same blood group, or they may differ in the amount of cholesterol (a type of fat) circulating in the blood.

Working back through the albums, consider those individuals who have passed away and the reasons for their deaths, such as cancer or heart disease. All these visible and invisible characters of an individual and also their medical histories are governed by their genes and the environments in which they have lived.

Genes are units of inheritance. Our characters — the structure and appearance of an individual, such as blue or brown eye colour — depend on the functions of genes. Genes also contribute to a person's behaviour and health, including susceptibility to certain diseases, such as heart disease. How do genes influence our individual characters and the type of disease each of us might develop? How are genes transmitted from generation to generation? Where are genes located? These are some of the questions that this course sets out to answer.

The following video clip provides a brief introduction to genetically inherited characteristics.

Video content is not available in this format.

You have selected this course almost certainly because you are interested in learning more about human genetics — the study of genes — and thus you may already be familiar with some of the scientific terms used. One of our aims in writing this course is to put you in a stronger position when you read about the science of genetics and listen to informed debates about modern techniques of manipulating genes, by increasing your understanding of the concepts and issues involved.

Many thousands of genes have been discovered, including many that have roles in disease. These genes are scattered throughout the human genome. But what is the human genome? The physical appearance of the bulk of the human genome is 46 long, thin structures known as chromosomes. The genome of all individuals (with a few exceptions) looks like that in Figure 1.1. It is along the length of each chromosome that the genes are located. The term genome is a combination of the two words ‘gene’ and ‘chromosome’.

Chromosomes are built up from a remarkable substance called DNA (which stands for deoxyribonucleic acid). DNA is breathtakingly simple in structure and yet capable of directing the way we grow, reproduce and survive; hence it is often referred to as the genetic blueprint — the plan — of human life. Since genes are part of chromosomes they too are composed of DNA, which is why DNA is referred to as the *genetic material*.

The following video clip contains a little more information about DNA.

Video content is not available in this format.

Figure 1.1The human genome: 46 long, thin chromosomes, made conspicuous by the use of dyes. This photograph shows chromosomes in a white blood cell, magnified approximately 1000 times.

Where, within the human body, are the chromosomes located? We, like many different types of organism, are composed of many millions of millions of cells; indeed, the cell is often referred to as the 'unit of life'. Cells are too small to be seen with the naked eye, but if you were to take a scraping from the inside of your cheek and look at it under a microscope you would see some cells, like the one in Figure 1.2. The important feature about cells for the human genome is the large structure called the nucleus, because it is inside the nucleus that the chromosomes are found. Within the nucleus of every cell in the human body (with a few exceptions) is a copy of the human genome. This adds up to a huge amount of DNA within any single individual.

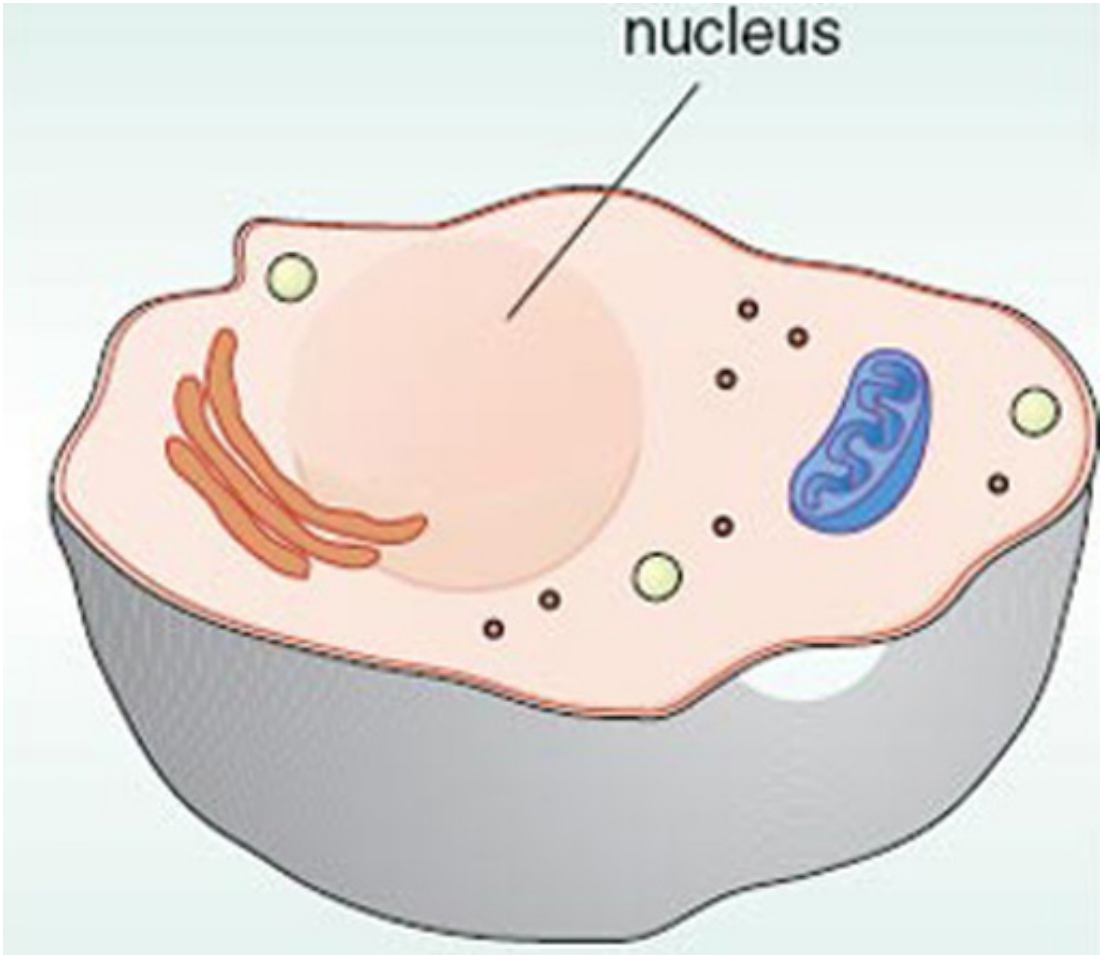

Figure 1.2 A drawing of a typical cell cut in two to reveal the nucleus. Note that about 20 such cells laid side by side would measure 11 mm on a ruler.

In order to understand more about genes and the rest of the genome, in 1991 a formal programme, the Human Genome Project, was established to discover all the genes along each chromosome and to sequence the entire length of the human genome. On Monday 26 June 2000, scientists announced world-wide that they had completed a rough or first draft of the sequence of the human genome. The powerful headlines shown in Figure 1.3 were the way that some British newspapers captured the attention of their readers, on or around that date. Is the Human Genome Project 'the breakthrough that changes everything' (Figure 1.3), and, if so, *how* will it? Does the draft sequence really have implications for 'every person on the planet' or just for scientists and doctors?

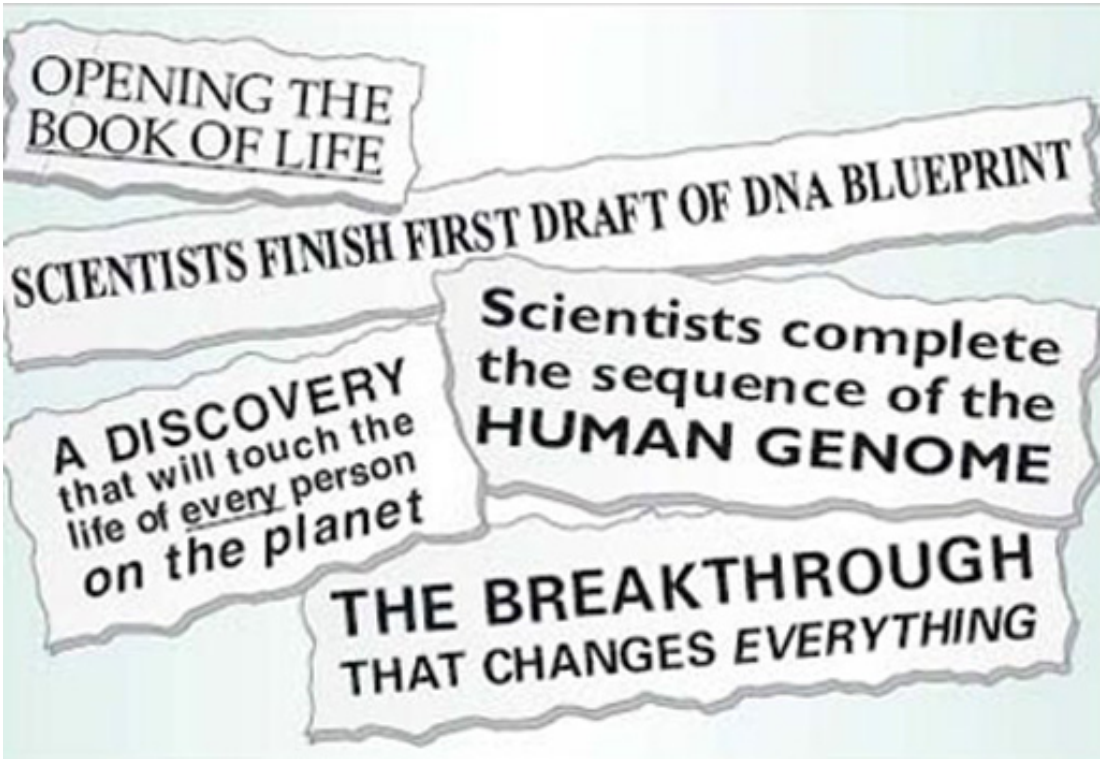

Figure 1.3 A collage of newspaper headlines, published on or around 26 June 2000, announcing the publication of the first draft sequence of the human genome.

What do the words in the headline 'sequence of the human genome' (Figure 1.3) mean? DNA contains information in a code that can be written in a four-letter language, A, C, G and T, in which each letter represents a different chemical (adenine, cytosine, guanine and thymine). Like all codes, the one in DNA carries information or instructions; in this case, ones that direct the growth and survival of each individual. Like every letter in each word in this sentence, to be meaningful, the letters A, C, G and T have to be in the correct order or sequence in each gene. The Human Genome Project involves identifying each of these letters in the correct sequence for each chromosome in turn for the whole genome.

Before we go any further, we should try to gain some appreciation of the immense scale of *one copy* of the human genome and hence the phenomenal scope of the task that faced the Human Genome Project. The human genome comprises approximately six billion, 6 000 000 000, letters (chemicals) of A, C, G and T, joined together in pairs, i.e. 3 000 000 000 pairs, in a linear sequence along the length of the chromosomes. In order to appreciate the scale of this, consider the following data. The typeface of a typical book enables approximately 5000 letters to be printed on one page. Thus the sequence of the human genome would take a total of 600 000 pages and fill about 3000 books. The project was made possible only by a large number of staff working in a substantial number of laboratories and, importantly, by database technology of computers. Each computer is linked to the internet, on which the results are published world-wide for free with no restrictions on their use or distribution.

By June 2000, the Human Genome Project had determined about 85% of the sequence of the four letters with about 99.9% accuracy. The figure of 85% was chosen as the point at which the genome would have scientific and medical value; hence the announcement on 26 June 2000 of the rough draft. By February 2001, about 90% of the sequence had been determined. But note that these drafts were far from perfect, containing many gaps. Since these initial publications, further sequencing has produced a more complete and accurate

sequence so that by 2003 very few gaps and errors remained in the then published sequence. Most of these uncertainties have now been resolved. The ultimate goals are to produce a completely finished sequence by filling in the remaining gaps and perfecting the accuracy, and to understand and interpret fully all the details of the genome sequence including the genes it contains.

Let's return to June 2000. Our understanding of the human genome did not begin on this date; in fact, we had been learning about it, bit by bit, throughout the whole of the 20th century. The study of genes began in 1900 when it was shown that genes govern inheritance in many different creatures. In 1907 it was shown that the same patterns of inheritance could account for the transmission of eye colour in humans. However, not until 1953 was the structure of DNA deduced by Watson and Crick. Hence, most of this course will consider aspects of genetics and the genome that were understood long before the publication of the first draft of the human genome.

Human genetics is a huge topic. It would be possible to write a whole course that focused on just one aspect of it. However, our goal in this course is to introduce not only the science, but also some of the health issues generated by the study of genes. The science deals with the structure and function of the genome within each individual and the medical aspects such as disease genes and gene therapy. Although there are thousands of genes, some with very complicated names and functions, we discuss only a few straightforward but representative examples in order to demonstrate the basic principles.

The health issues include, for example, DNA testing for the presence of genes related to specific diseases. An issue is a topic that can always be considered from more than one point of view or perspective. For example, if the fetus of an expectant mother tested positive for a debilitating genetic disease, she might be confronted with the choice of a termination or having an affected child. The expectant mother's point of view and her partner's might be very different from that of the medical profession or society in general. Hence, health issues, in turn, raise social and ethical issues.

1.2.1 The human genome: chromosomes and genes

Almost all of the different types of cell in an individual contain the same genetic material in the chromosomes, as a consequence of mitosis. You had a glimpse of the appearance of the 46 human chromosomes in Figure 1.1, and in this course we examine their size, shape and genetic composition.

Chromosomes can be regarded as strings of genes arranged along their length, rather like beads on a thread. Each gene consists of a short section of the DNA that runs the full length of each chromosome. Genes direct our characters, such as eye colour, and also the processes that go on inside us, from the small-scale processes of cell division to the large-scale processes of development and reproduction.

1.2 2 The chromosomes that constitute the human genome

Laboratory techniques are available to aid in the preparation and staining of the chromosomes from a single cell, so that they are readily distinguished and can be photographed under the microscope. During mitosis, the chromosomes become visible (because they have condensed) and it is during mitosis that chromosome number, size and shape can be most easily studied. Every *species* has a particular number of chromosomes, each with a characteristic size and shape. For example, chimpanzee cells have 48 chromosomes, turkey cells have 82, and the cells of some species of ferns have over 1000 chromosomes!

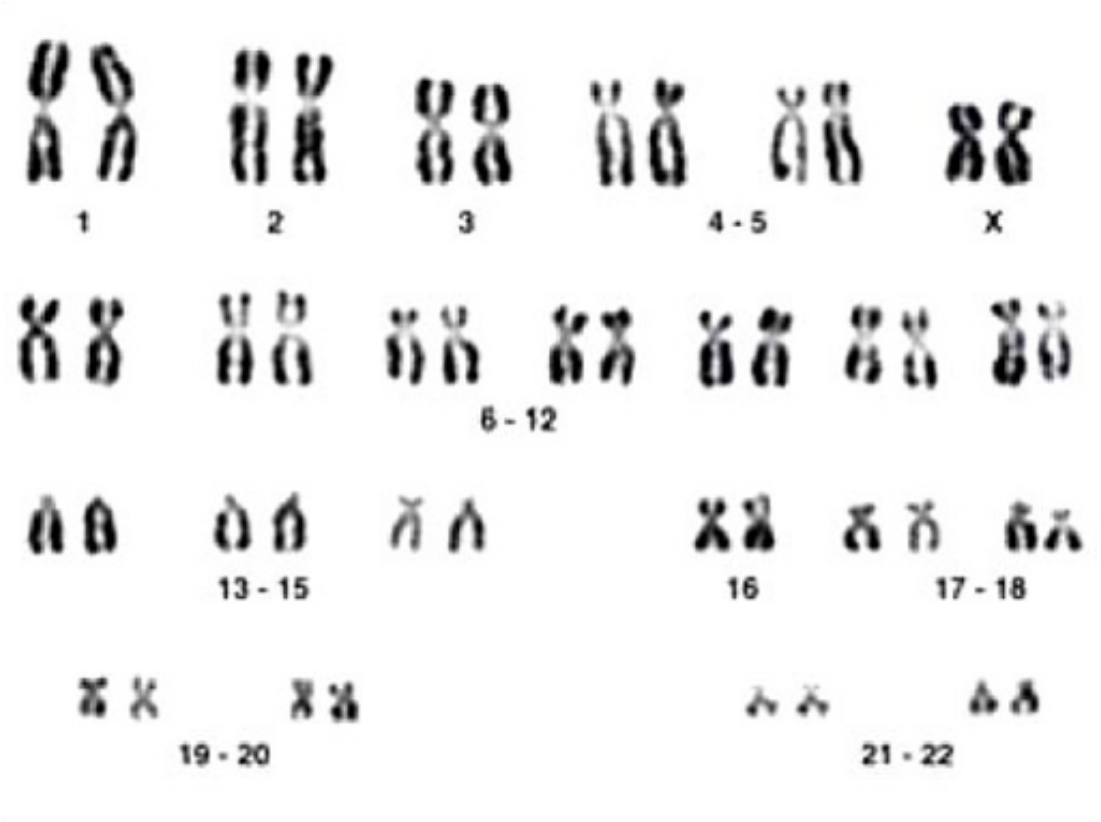

Figure 2. Photograph of the stained chromosomes of a human female, arranged as a karyotype (photographed through a microscope, magnified approximately 1000 times). The chromosome images are cut from a photograph of a spread of chromosomes at mitosis (such as the one shown in Figure 1.1). The pairs of chromosomes have been arranged in a conventional sequence and are numbered 1 to 22, with one pair of X chromosomes. Each chromosome has duplicated before mitosis begins, and so consists of two chromatids about to separate.

Figure 2.1 shows the chromosomes of a human female, arranged to reveal their features. This distinctive pattern is described as the human female **karyotype**, that is, the pattern of chromosomes that is unique to human females. The same term, karyotype, is used for the standard chromosome set of an individual, as in 'human female karyotype', or of a species, as in 'human karyotype'. (Each chromosome in Figure 2.1, consists of two chromatids about to separate, one into each of two new cells.)

SAQ 1

What is the most striking feature of the human chromosomes shown in Figure 2.1?

Answer

The 46 chromosomes are present as 23 pairs of various sizes; the members of each pair look the same.

The chromosomes in 22 of the pairs have been given a number starting with the largest and progressing to the smallest. Thus, there is a pair of chromosome 1, a pair of chromosome 2, etc. Members of a pair are said to be *homologous*, which means 'having a similar structure', thus there are 22 **homologous** pairs of chromosomes. Although the chromosomes of some homologous pairs are longer or shorter than others, it is still

difficult to distinguish between several pairs of chromosomes on this criterion alone. For example, look at the pairs of chromosomes numbered 6 to 12 in Figure 2.1: if those chromosomes were mixed up it would be difficult to separate them into pairs again on the basis of visual appearance alone. This difficulty was overcome in the 1970s by the discovery of special staining techniques that produce distinctive patterns of bands on chromosomes (Figure 2.2).

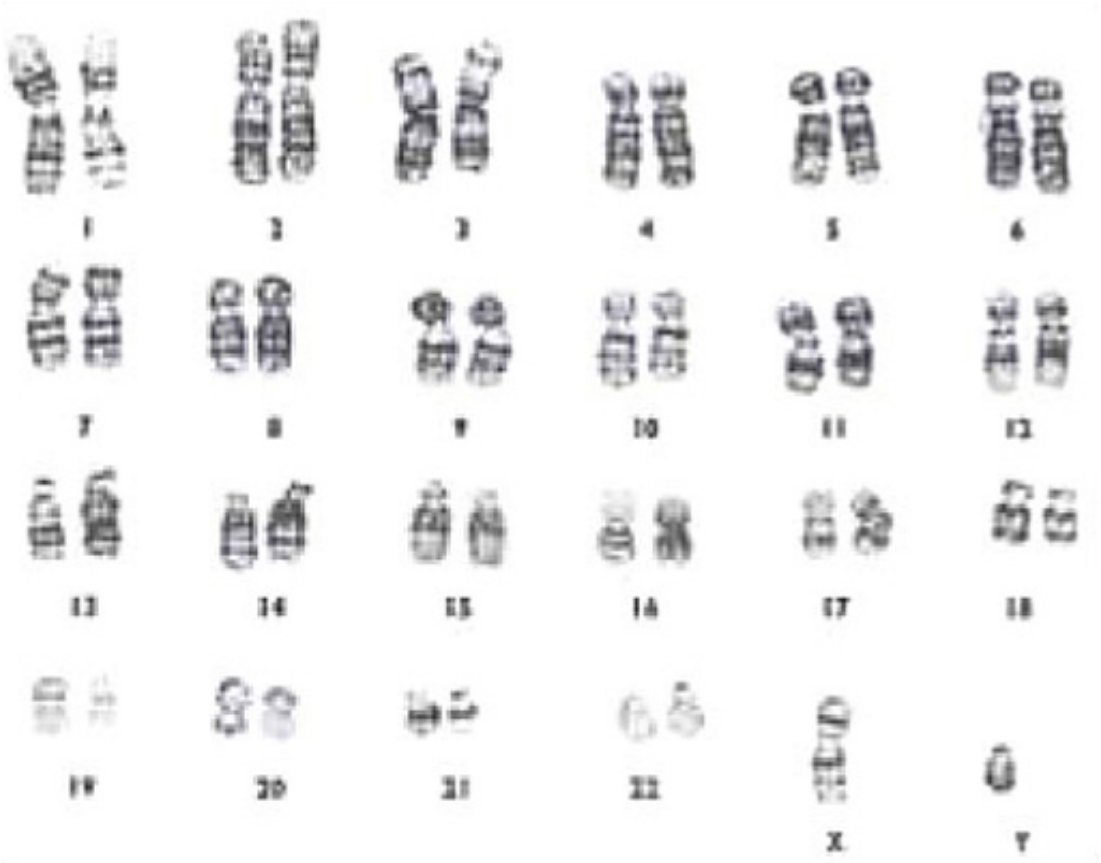

Figure 2.2 Photograph of the chromosomes of a human male, arranged as a karyotype and stained to reveal their characteristic banding pattern (photographed through a light microscope, magnified approximately 1000 times).

Such techniques have made it possible to define the differences between the members of one pair and those of another, as shown in Figure 2.2. The chromosomes of a human male, arranged as a karyotype, could be from any cell in the body (with a few exceptions).

SAQ 2

How does the human female karyotype (Figure 2.1) differ from the human male karyotype (Figure 2.2)?

Answer

The female karyotype has two X chromosomes whereas the male has one X chromosome and a small Y chromosome.

The X and Y chromosomes are called **sex chromosomes** because they play an important role in sex determination: females with a pair of homologous X chromosomes are described as XX, and males are described as XY. Apart from these sex

chromosomes, both males and females contain similar sets of 22 homologous pairs of non-sex chromosomes, or *autosomes*.

SAQ 3

In Figure 2.1, what does the banding pattern of the homologous pairs of chromosomes numbered 6–12 reveal?

Answer

First, each pair can be distinguished from all other pairs, and second, each chromosome appears to have an identical banding pattern with its partner.

The two observations that you have just made about chromosomes reveal important information about inheritance. We will look at the implications of each of these observations in turn.

Each of the 24 *different* kinds of chromosome that may occur in a human cell (that is, chromosomes 1 to 22, plus X and Y) carries different *genes*; each has particular genes arranged in a specific order along its length. However, both partners in a pair of homologous chromosomes, for example the pair of chromosome 1, carry the *same* genes in the *same* order. This means that each gene along the 22 autosomes, and the X chromosomes in females, is present twice in the nucleus of almost all cells, a feature that has important consequences for inheritance. Furthermore, the set of genes is the same in all humans.

Different chromosomes contain different genes, but the partners in a pair of homologous chromosomes carry the same genes in the same order along their lengths.

Figure 2.3 shows that the two sex chromosomes look quite different from each other. They also carry different genes and, given the small size of the Y chromosome, you may not be surprised to learn that it contains very few genes, the most important of which is the gene that carries instructional information for the development of *testes* (male reproductive glands; singular testis) rather than *ovaries* (female reproductive glands; singular ovary).

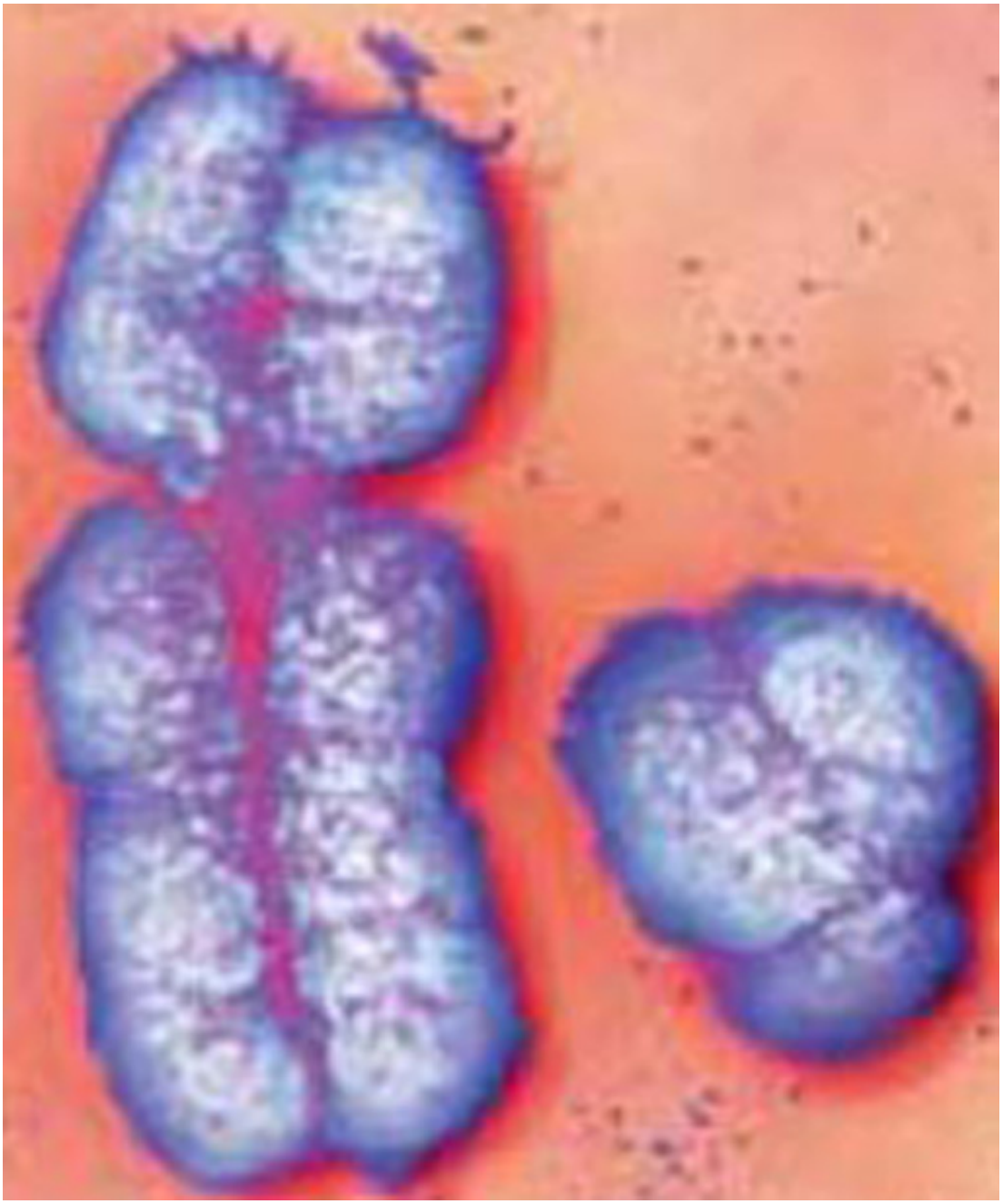

Figure 3.3 A pair of male sex chromosomes: the X chromosome (on the left) and the Y chromosome (on the right). Each chromosome consists of two chromatids attached at the centromere. This photograph was taken with the aid of a scanning electron microscope.

1.2.3 The transmission of genetic material

The full complement of 46 chromosomes in the human genome, the *diploid* number, is restored at fertilization. As Figure 3.1 shows, all the somatic cells and cells in the testes and ovaries arise from the same fertilized egg by the process of mitosis; the cells all contain copies of the same genetic material (with some exceptions).

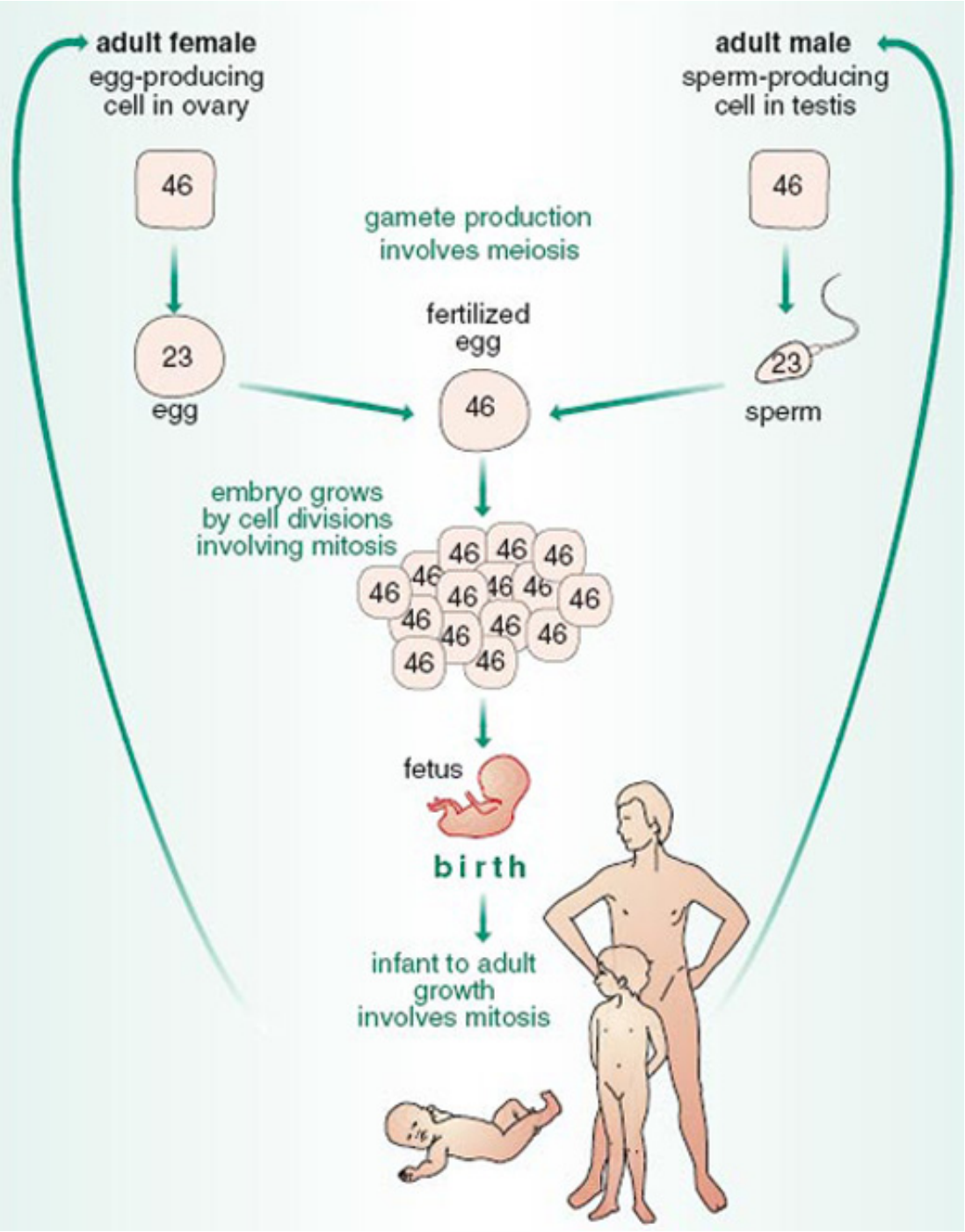

Figure 3.1 The changes in chromosome number in the human life cycle. Gamete cells contain 23 chromosomes, whereas all other cells of the body (with the odd exception) contain 46 chromosomes. We have omitted the nuclear membrane in each cell for simplicity, and we continue to do this in most subsequent figures.

Activity 1

From Figure 3.1, do the gamete-producing cells have the same chromosome number as the somatic cells?

Answer

Yes; they have 46 chromosomes, the same number as somatic cells.

Each gamete-producing cell contains an identical copy of the genetic material in the somatic cells. Only the gametes contain half the chromosome number.

Now look at Figure 3.2, which illustrates the production of gametes in each parent and the possible combining of gametes at fertilization with reference to the sex chromosomes only. We have omitted all the other chromosomes to make it easier to follow the sex chromosomes. First, we consider the production of gametes, and in the next section we examine fertilization. Each one of the egg-producing cells in the ovaries of the female contains two X chromosomes, whereas the equivalent cells in the male each contain one X chromosome and one Y chromosome (top row of Figure 3.2). In the male, the X and Y chromosomes separate from each other during meiosis, the X chromosome to one sperm and the Y chromosome to another. Similarly, in the female, the two X chromosomes separate from one another, each one going into a different egg cell.

Activity 2

On the basis of the middle row of Figure 3.2, what are the proportions of X-bearing and Y-bearing gametes produced by the male?

Answer

Half of the gametes are X-bearing gametes and half are Y-bearing.

This answer can be expressed as a ratio in a number of ways; these are described in Box 3.1, *Ratios*.

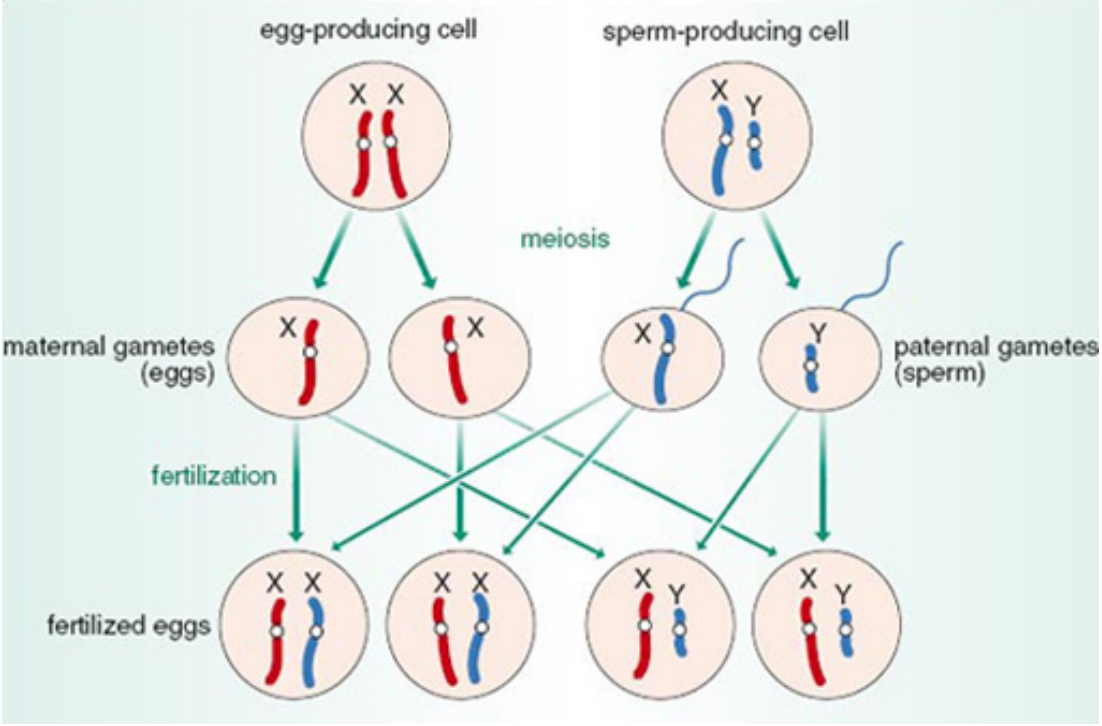

Figure 3.2 The distribution of human sex chromosomes during gamete formation and at fertilization. Note that the chromosomes are drawn as single structures in order to make it easier to follow the events.

Box 3.1 Ratios

Ratios are another way of expressing proportions. Ratios are usually written as two numbers separated by a colon (:). As the answer to the in-text question preceding this box shows, X- and Y-bearing gametes are produced by the male in equal numbers, so the ratio of X-bearing gametes to Y-bearing gametes is one to one, written as 1 : 1.

We are also interested in the proportion of the total number of gametes that are X-bearing gametes. The male produces two different types of gamete, of which one is X-bearing; so the ratio of X-bearing gametes to the total number of male gametes is 1 : 2.

Although not relevant in this case, it is important to note that ratios are usually simplified to the smallest whole numbers by dividing both sides by a common factor. For example, in a class of 4 boys and 8 girls, the ratio of boys to girls is 4 : 8, which is the same as 1 : 2, where both the numbers have been divided by 4.

You need to be careful about which ratio is being considered. Consider a practical example of a group of 20 people, 6 of whom have red hair and 14 of whom do not. The ratio of red-haired people to the total number of people is 6 : 20 (or 3 : 10). But now consider the ratio of red-haired people to non-red-haired people in the group: it is 6 : 14 (or 3 : 7).

SAQ 4

Suppose that 2 out of 10 people in the UK have blue eyes. What is the ratio of people with blue eyes to those who don't have blue eyes?

Answer

The ratio is 2 : 8 (or 1 : 4).

Did you fall into the trap and answer 2 : 10 (or 1 : 5)? This is the ratio of people who have blue eyes to the *total* number of people. If we had asked what is the ratio of people who don't have blue eyes to those who do, the answer would have been 8 : 2 (or 4 : 1). So always read the question carefully and make sure when expressing ratios that it is clear what each number refers to.

SAQ 5

Question 3.1, A class of children consists of 10 boys and 15 girls. What is the ratio of girls to boys?

Answer

The raion is 3:2 (i.e.15 girls to 10 boys); note that we simplified the numbers.

The behaviour of the sex chromosomes during meiosis is similar to that of all other pairs of chromosomes, that is the number is halved in each gamete. Thus the members of each of the 22 homologous pairs of autosomes in a gamete-producing cell also separate from each other during meiosis; one member of each chromosome pair enters each gamete. A consequence of the separation, or **segregation**, of members of homologous pairs of chromosomes (or the X and Y chromosomes) is that each gamete contains one member of each pair of autosomes and one sex chromosome (either X or Y).

We can now link the description of gamete production at the cellular level with the stages of meiosis. In males, meiosis begins at puberty and continues well into old age; each meiotic division takes just three hours. In contrast, in females the process of meiosis in an egg-producing cell *begins* before birth. But the halving of chromosomes during meiosis is halted within the egg-producing cell and is completed some time between puberty and menopause. Thus the process of meiosis and egg production in females is halted for many years before being completed. During a woman's reproductive life, one cell normally completes the meiotic division and gives rise to one egg during each menstrual cycle. The process of meiosis resulting in the production of an egg shortly after puberty has been halted for 10–15 years, and the process of meiosis resulting in the production of an egg just before menopause will have been halted for about 50 years!

1.2.4 Fertilization

Now that we have considered the production of gamete cells, including the process of meiosis, we can examine the next stage of reproduction, the process of fertilization, which occurs inside the female's reproductive tract. As fertilization occurs, the successful sperm stops swimming and a change takes place in the egg cell membrane, which prevents any other sperm from fusing with it. The nucleus of the sperm cell is injected into the cytoplasm of the egg cell. The chromosomes of the fertilized egg thus consist of one set from the egg and one set from the sperm.

The following video clip shows the process of fertilization in more detail.

Video content is not available in this format.

We have seen that fertilization restores the number of chromosomes to 46 (Figure 4.1)

- Why is meiosis such an important feature of reproduction?
- If the chromosome number were not halved in the gametes prior to fertilization, the fertilized egg would contain 92 chromosomes, twice the normal number in the parents' cells.

The fertilized egg grows and develops into an embryo by repeated mitotic divisions. The embryo in turn grows and develops into a fetus, baby, child and finally an adult. The life cycle shown in Figure 4.1 depicts all of the stages that a human must go through from conception through birth to reach adulthood.

One consequence of fertilization can be examined by following the behaviour of the sex chromosomes. We can do this by returning to Figure 4.1, the top part of which we considered earlier. Figure 4.2 is a **mating diagram**, which shows the parents' gamete-producing cells, the gametes and the possible combinations of sex chromosomes at fertilization. An individual with a Y chromosome will develop into a male.

- From Figure 4.2, what is the expected ratio of female offspring to male offspring?
- Half of the sperm contain an X chromosome and every egg also contains one. Therefore, half of the fertilized eggs will be XX, and half will be XY, so the ratio of female to male offspring is 1 : 1.

The ratio of females to the total number of offspring is 1 : 2. Thus, for any fertilization, the *chance* of producing a girl is 1 in 2, and the chance of producing a boy is the same, 1 in 2. The process of meiosis, which governs the separation, or segregation, of X and Y chromosomes during gamete formation, not only accounts for the occurrence of XX

(female) and XY (male) individuals, but is also responsible for the production of the two sexes in the ratio of approximately 1 : 1 in any population.

Figure 4.3 represents a different way of showing the same information in Figure 4.2. Either way of presenting the details of a mating can be used, and when producing your own mating diagrams you can choose whichever is easier for you. In Figure 4.3, the fertilizations between the various combinations of gametes are shown in boxes. Along the top you can see the two types of gamete, X and Y, produced by a male; down the left-hand side you can see the two gametes produced by a female. Inside the other boxes are the products of fertilization between the various gametes. Thus, for example, the top right-hand box records the outcome of the fertilization of an egg with a Y-bearing sperm. An examination of the four boxes should convince you that the expected ratio of XX individuals to XY individuals is 2 : 2, which is the same as 1 : 1.

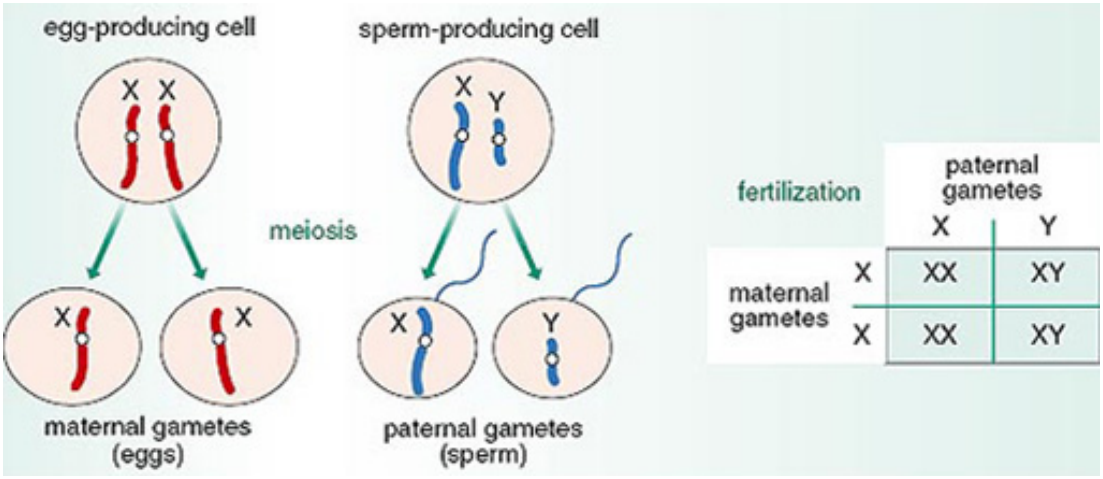

Figure 4.3 An alternative way of laying out the matings shown in Figure 4.2

This ratio of 1 : 1 follows for two reasons. First, the two different gametes, X and Y, are produced in equal numbers in sperm. This is a consequence of segregation of the two sex chromosomes. Second, fertilization is *random;* that is, an egg is equally likely to be fertilized by an X-bearing sperm as by a Y-bearing sperm.

The 1 : 1 ratio of males to females is a consequence of:

segregation of chromosomes during meiosis into different gametes in equal numbers;

the combining of a female gamete and a male gamete at fertilization occurring at random.

Conclusion

This free course provided an introduction to studying Science. It took you through a series of exercises designed to develop your approach to study and learning at a distance and helped to improve your confidence as an independent learner.

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