Unit
2B

Chapter 22
Making informed choices

Figure 22.1 The choices we make, such as vaccination for children, can have profound effects on the health and well-being of individuals and of society in general

Unit content

The relevance of human biology to everyday life
The rate of change in human biology means that there is a range of alternative treatments available. Each treatment has its risks, ethical concerns and benefits based on individual variations and the condition being treated. Health choices can be based on myths or misconceptions about human biology.

Medical technologies:

- sex selection of embryo to avoid genetic disease
- treatment for various genetic diseases.

Health choices:

- performance enhancing *e.g. steroid use*
- parents' choice for infants *e.g. diet and immunisation choices*.

Inheritance
Mutations affect cellular and body functions. Genetic counselling uses information from pedigrees, genetic testing to provide an analysis of the risk associated with some of these mutations.

Genetic testing of parents and offspring for:

- gene and chromosomal abnormalities.

Human Genome Project:

- information provided by the Human Genome Project
- range of possible uses for this information.

This chapter considers the relationship between some aspects of human biological knowledge and the impact of that knowledge on you and on society. As knowledge about humans expands, and as technology develops, there are implications for the choices we make, for our attitudes and values, and for the attitudes and values of society in general. Making appropriate choices can positively affect your health and wellbeing. A good decision is one that is well informed. In this chapter, information is presented to help you make appropriate choices that may lead to a healthier, happier and longer life.

Sex selection

In Australia, in 2006, 1057 boys were born for every 1000 girls. Is there anything that couples can do to ensure that their baby is a boy or a girl?

Sex selection means choosing to have a baby of a desired sex. Various natural methods have been promoted to increase the chances of a couple having a baby of a particular gender. Most of these methods involve the timing of sexual intercourse in relation to ovulation. Some also involve diet or the positions adopted by the couple during sex. There is no reliable evidence that any of these natural methods have had any influence on the sex of the baby.

Reliable methods of sex selection all involve considerable use of technology and considerable financial cost to the parents.

Pre-implantation methods

Sperm sorting involves separating sperm with an *X* chromosome from those with a *Y* chromosome. The sorted sperm can then be used either for artificial insemination or for in-vitro fertilisation (IVF). Sorting techniques are not completely accurate so the method is only 65% to 85% effective.

Pre-implantation genetic diagnosis (PGD) is where embryos resulting from IVF are tested to see whether the cells have an *X* or *Y* chromosome. An embryo of the desired sex can then be implanted into the mother's uterus. Selection of an embryo on the basis of sex is illegal in some Australian states.

Post-implantation methods

Ultrasound and/or amniocentesis can be used to determine the sex of a foetus after 16 to 20 weeks gestation. An abortion could then be performed if the baby is not of the desired sex. This raises all of the ethical issues relating to abortion that were discussed in Chapter 16. There is also a significant risk to the mother when a pregnancy is terminated after four to five months.

Genetic diseases

Genetic diseases (often called **genetic disorders**) are caused by abnormalities in a person's genes. Each gene has the code for manufacture of a particular protein (see Chapter 17) and if the gene is faulty then the protein produced will not carry out its normal function.

There are four different types of genetic disorders.

1. **Single gene disorders** (also called **Mendelian** or **monogenic**) result from a change (mutation) in the DNA of just one gene. Such disorders are inherited

in recognisable patterns so that by studying family history the probability of their occurrence can be predicted. Examples of single gene disorders are cystic fibrosis, Huntington disease, haemophilia and sickle cell anaemia (see Chapters 18 and 19).

2. **Polygenic disorders** (also called **multifactorial** or **complex**) are caused by mutations in a number of genes and interaction with environmental factors. These disorders do run in families but because of the number of influences involved, prediction of their occurrence is very difficult. For example genes that influence susceptibility to breast cancer have been located on six different chromosomes. Other examples of polygenic disorders are Alzheimer's disease, diabetes, arthritis, heart disease and high blood pressure.
3. **Chromosomal disorders** result from missing chromosomes, extra copies of chromosomes or cases where chromosomes have broken and rejoined. Down syndrome, trisomy 21, is one such disorder where the affected individual has three copies of chromosome 21 (refer to Chapters 20 and 21).
4. **Mitochondrial disorders** are rare and are caused by mutations in the mitochondrial DNA (see Chapter 17).

Prevention of genetic diseases

At present there are no effective cures for genetic conditions so prevention is the best choice. The only way to prevent a genetic disease is to prevent the birth of an affected foetus. Two possibilities are available to parents who are at risk of having a child with a genetic disorder. Both options involve parents making difficult choices. Genetic counsellors are often employed to advise couples on the risks and options available.

Reproductive choice

In the case of single gene disorders, knowing the pattern of inheritance enables the risk of a couple having an affected child to be determined. Also, if the parents already have a child affected by a recessive single gene disorder then the chances of any subsequent child being affected is 1 in 4. Knowing the odds, the couple must then choose between having another child and not having further children.

Prenatal diagnosis

If there is a risk of a baby having a genetic abnormality, samples of the baby's cells can be taken using amniocentesis or chorionic villus sampling (see Chapter 15). The baby's chromosomes or DNA can then be analysed. Should a genetic problem be diagnosed the parents then have to decide whether to continue with the pregnancy with the knowledge that the child has a genetic abnormality or to terminate the pregnancy. These are difficult decisions to make.

For further information on genetic diseases go to:
- http://www.biotechnologyonline.gov.au/human/gendisorders.cfm
- http://www.ornl.gov/sci/techresources/Human_Genome/medicine/assist.shtml

Sex selection

Where there has been a history of a sex-linked disorder in a family, selecting the sex of an embryo, either before or after implantation, could be appropriate. If pedigree analysis shows that the mother is a carrier for an X-linked recessive allele there is a 50% chance that a male child will inherit that allele. Selection of a female embryo would therefore avoid the possibility of an affected child.

Treatment of genetic diseases

Prevention of symptoms

For some genetic disorders treatments are available that will prevent symptoms from developing if treatment is applied early enough. This means that the condition must be diagnosed before any symptoms appear.

One of the best known examples of preventive therapy is for phenylketonuria (PKU), a metabolic disorder that can cause severe mental retardation. In Australia all newborn babies are tested for PKU by taking a drop of blood from a prick in the heel. The blood is placed on a piece of absorbent paper, called a Guthrie card, and the dried blood can be examined later and then stored indefinitely. If a baby is diagnosed with PKU it can be placed on a diet that is very low in the amino acid phenylalanine and intellectual development will not be affected.

A baby's blood sample can also be tested for hypothyroidism, another condition that can cause intellectual disability and growth problems. Early detection and treatment enables children to develop normally.

Replacement therapy

Replacement therapy is where the protein that is missing or defective as a result of a faulty gene is replaced with a functioning protein. Examples are diabetes and growth hormone deficiency. Patients with type 1 diabetes are given regular injections of the hormone insulin, the protein that is missing as a result of a genetic fault. In the same way people suffering from growth hormone deficiency are given injections of growth hormone.

Gene therapy

Gene therapy aims to treat or cure genetic abnormalities by replacing faulty genes with healthy ones. Around 4000 genetic disorders are known and more are being discovered. As a result of the Human Genome Project (see Chapter 19) the location of all of these potentially faulty genes is known and replacement of the genes becomes a possibility. Gene therapy is a major area of investigation into the treatment and cure of disease but it is still in the trial stages.

Germ line therapy involves replacing the defective gene in either a sperm or an egg. This means that the changed gene would be passed on to future generations. Germ line therapy has not been attempted in humans for technical and ethical reasons. It is technically very difficult and it raises many ethical considerations because it involves changing the human genome. Many countries have banned germ line therapy and in Australia the guidelines are so strict that they almost amount to a ban.

Somatic cell gene therapy is where functional genes are inserted into non-reproductive cells of the body. Such genes, inserted into bone, muscle, skin, brain or other cells, are not passed on to offspring, but improve the health of the recipient.

There are two ways in which healthy genes are transferred into body cells.

In ***ex vivo*** (meaning outside the body) gene transfer the genes are transferred into cells grown in culture. The modified cells are then introduced into the patient (Fig. 22.2). This method can only be used for tissues where cells can be removed without harm to the patient and where they will grow and survive for long periods of time after return to the person.

In ***in vivo*** (inside the body) gene transfer the replacement genes are transferred directly into the tissues of the patient. This may be the only option for tissues such as brain cells that cannot be cultured in large numbers.

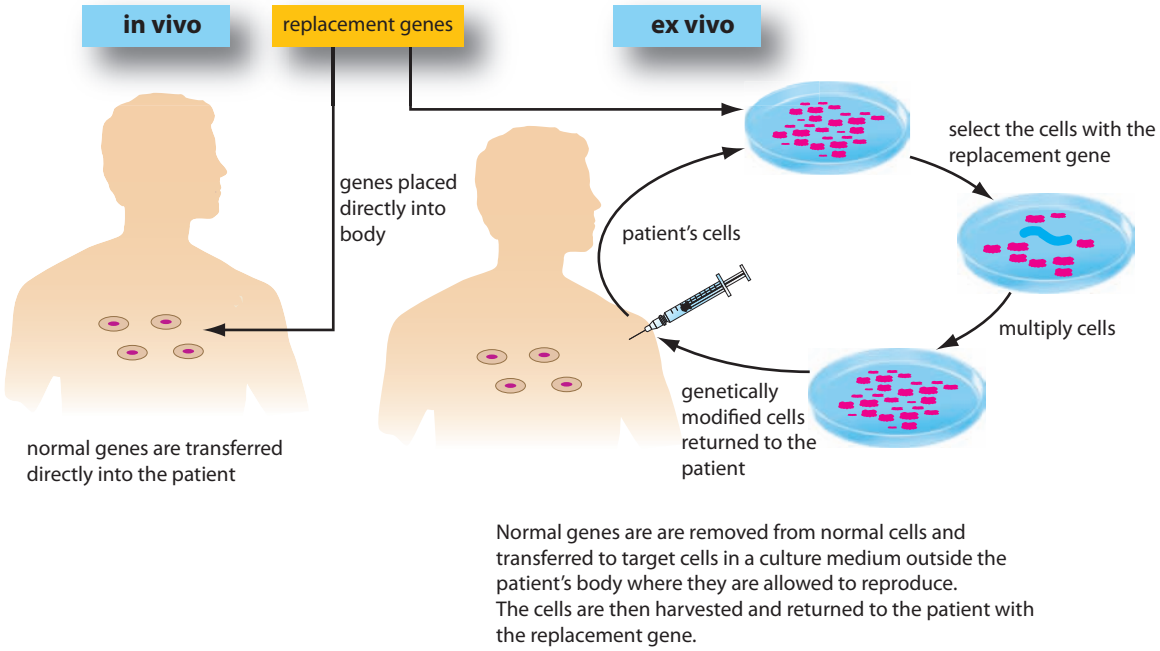


Figure 22.2 Ex vivo and in vivo methods of gene transfer

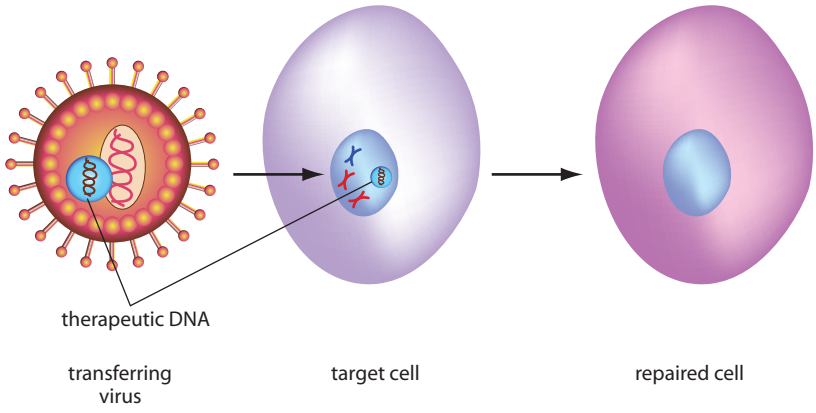
Introduction of the appropriate genes into cells is the crucial part of gene therapy. Viruses have evolved to infect cells as they reproduce by delivering their genetic material into the infected cells. By removing any disease-causing genes from a virus, and inserting the replacement genes to correct the patient's disorder, viruses can then be used to transfer those genes into a patient's cells (Fig. 22.3). If the replacement genes can be delivered into stem cells, then all the new cells produced as a result of mitosis of those stem cells will contain the replacement gene. This method has been used successfully in trials but a problem is that the patient's immune system eventually destroys the virus.

Several non-viral means of gene transfer have been tried. The normal DNA (therapeutic DNA) can be directly injected into cells. This method requires large amounts of DNA and can only be used for certain tissues.

Another approach is to include the DNA in **liposomes**—lipid spheres with a watery core. The liposomes fuse with the target cell membranes and are then able to pass the DNA through the membranes into the target cells.

Yet another line of research is to try to introduce an artificial human chromosome into target cells—chromosome number 47. Such a chromosome would exist alongside the other 46 and would not affect their workings. Each time the cell divided by mitosis the extra chromosome would be faithfully replicated along with the others. The additional chromosome could carry a large number of genes but it would be very difficult to deliver such a large molecule into the nucleus of a cell.

Figure 22.3 Using a virus to transfer genes into a patient's cells



Nutrition in infancy

A major decision a mother must make is whether she will breastfeed or bottle-feed her baby. There are advantages and disadvantages to each method but, in general, breastfeeding a child is more natural and tends to develop a close mother–child bond.

Breastfeeding

Human milk is the perfect food for the developing infant (Table 22.1). It meets all the nutritional needs of an infant for the first six months of life and can be an important food for a further six months. Breast milk provides protein, fat and carbohydrate at levels unique to human requirements. It is thus ideal for the development of babies and infants. Furthermore, the levels of nutrients can vary from feed to feed because the mother's metabolism is able to produce milk appropriate for her baby at a particular time.

Commercial preparations, based on cow's milk, are available for mothers who cannot (or who choose not to) breastfeed. These formula milks resemble breast milk as closely as possible but, despite great improvements in quality over the past 20 years, they still do not have the unique properties of breast milk.

When a baby is first born the milk secreted is a watery, yellowish-white fluid called colostrum. **Colostrum** has a similar composition to milk, but contains little or no fat and has a high content of the mother's antibodies. These antibodies can be absorbed through the infant's intestine to give temporary immunity to those diseases to which the mother is immune.

Table 22.1 The composition of milk

Component	Approximate concentration (g/100 mL)		
	Colostrum	Human milk	Cow's milk
Water	89.4	89.7	87.0
Lactose (milk sugar)	5.3	7.4	4.8
Lipid	2.9	4.2	3.7
Protein	2.7	1.3	3.3
Minerals			
Potassium	0.074	0.060	0.138
Calcium	0.031	0.035	0.044
Phosphorus	0.014	0.015	0.096
Sodium	0.048	0.015	0.058
Iron	90 µg	76 µg	100 µg
Vitamins			
A	108 µg	60 µg	40 µg
C	0.004	0.004	0.001
D	–	0.01 µg	0.06 µg

(Note: µg = microgram; 1 000 000 µg = 1 g.)

Breastfeeding has many important advantages:

- Breast milk is sterile.
- Breast milk is always at the correct temperature for the infant.
- Breastfed babies are not subject to mineral imbalance.
- Obesity is rare in breastfed babies.
- Breast milk contains antibodies, produced by the mother, which afford the infant some protection against infections.
- Breastfeeding is associated with decreased risk of diabetes and sudden infant death syndrome.
- The act of breastfeeding builds a strong bond between child and mother.
- The suckling stimulus on the nipple causes the release of hormones, which help to shrink the mother's uterus—much enlarged during pregnancy.
- Mothers who breastfeed are less likely to suffer ovarian or breast cancer, or bone loss later in life.
- Breastfeeding is more economical than purchasing milk formulae.

Although the amount of iron in breast milk is small (see Table 22.1), the iron is more efficiently absorbed than iron in formula milks. In any case babies are born with sufficient stores of iron for the first six months of life. Once the baby is introduced to solid food it is important that iron-rich foods are included.

While the mother is breastfeeding, ovulation may be suppressed by the high level of the hormone prolactin. However, pregnancy can occur, and breastfeeding does not constitute a reliable method of contraception.

Infant vaccination

The World Health Organization (WHO) rates clean water supplies and the introduction of vaccines as the two public health measures that have had the greatest impact on people's health. **Vaccination** means giving a person a substance, a **vaccine**, that makes the immune system respond so that the person is less likely to be infected by a particular disease. This results in the person being 'immunised'. **Immunisation** can also occur naturally; people who catch an infectious disease and recover are unlikely to catch the disease again. Thus, there is a slight difference in the meaning between vaccination and immunisation, but the two words tend to be used interchangeably.

One of the important choices that parents must make is whether to have their children vaccinated in infancy.

Immunity

Immunity is resistance to infection by invading micro-organisms. When a foreign substance enters the body it triggers an **immune response**. Substances that trigger such responses are called **antigens**. Antigens are large molecules. They may be proteins, carbohydrates, lipids or nucleic acids. An antigen could be a whole micro-organism, such as a virus particle or bacterial cell, or it could be part of a bacterium, such as the cell wall or outside capsule. Toxins produced by bacteria are also antigens. Antigens are not necessarily associated with micro-organisms. Tissues transplanted from another person, blood cells of a foreign blood group, and such things as pollen grains and egg white contain antigens.

In Australia most people are vaccinated against the diseases for which vaccines are available. Childhood vaccination is not compulsory but 90% of infants have been vaccinated by 12 months. The recommended vaccination schedule for Australians is shown in Table 22.2.

Table 22.2 Recommended vaccination schedule for Australians

Age	Recommended vaccination
Birth	Hepatitis B
2 and 4 months	Diphtheria, tetanus, whooping cough (DTP); polio; hepatitis B; *Haemophilus influenzae* type B (HiB); rotavirus*; pneumococcal**
6 months	Diphtheria, tetanus, whooping cough, polio, HiB, rotavirus, pneumococcal
12 months	Measles, mumps, rubella (MMR); hepatitis B; HiB, meningococcal C***
18 months	Chickenpox
4 years	Diphtheria, tetanus, whooping cough; polio; measles, mumps, rubella
10–13 years	Hepatitis B, chickenpox
15–17 years	Diphtheria, tetanus, whooping cough
50 years and over	Influenza (annually), pneumococcal
65 years and over	Influenza (annually), pneumococcal

Source: NHMRC Australian Standard Vaccination Schedule 2007
* Protects against a highly infectious disease of the small intestine; most cases occur in children under 5 years.
** Protects against a bacterial infection of the lung that may lead to pneumonia if it occurs in children or the elderly.
*** Protects against a bacterial infection of the membranes around the brain.

Herd immunity

Due to the high levels of vaccination, Australia has not had a case of wild polio virus since 1978, there have been no cases of diphtheria since 1993, and tetanus is very rare.

As the incidence of infectious diseases declines, people become complacent and may consider that the risk of side effects from the vaccine is higher than the risk of contracting the disease. If vaccination rates decline, a serious outbreak of a disease could occur. This happened in the United Kingdom in the 1970s when large outbreaks of whooping cough occurred.

The immunity of the population, known as **herd immunity**, depends on a high proportion of individuals being immunised. A disease can only spread from an infected person if there is an unimmunised person available to catch it. Thus, it can be argued that having yourself and your children immunised is a social responsibility as it is contributing to the herd immunity.

Other health choices

Both males and females need to make choices that improve their chances of maintaining health. Men's health has become an issue in recent times because studies have shown that men are much more likely than women to neglect their health. It has been found that men tend to ignore, or not recognise, symptoms of ill health and they delay a visit to a doctor. Women on the other hand are more likely to acknowledge the symptoms and take action sooner. These are generalisations of course but health authorities have taken them seriously enough to mount education campaigns aimed at improving men's awareness of health issues and encouraging men to make appropriate choices earlier.

Enhancing performance

Many dietary supplements and sports drinks are advertised as performance improvers. Their use is legal and consumption at moderate levels is unlikely to harm a person's

health. However, there is little evidence that dietary supplements and sports drinks do actually improve performance.

Performance enhancing substances are most often taken by athletes trying to gain an advantage in their particular sports. The World Anti-Doping Agency (WADA) develops and implements uniform worldwide standards relating to the use of drugs in sport. These standards attempt to ensure that all athletes compete on an equal footing and that athletes do not have to risk their health to attain a high standard of performance.

Although many prohibited drugs are able to increase athletic performance they do have side effects that can cause death or life-long disability.

Anabolic steroids

Anabolic steroids are a group of prohibited drugs that have been widely used to improve athletic performance. They stimulate protein synthesis, especially in skeletal muscles and may be prescribed to treat various illnesses but their use without a doctor's prescription is illegal. Anabolic steroids include the male sex hormone testosterone and synthetic substances that have similar effects. In addition to increasing muscle and bone growth they also increase the body's male characteristics. They are therefore sometimes known as **anabolic androgenic steroids (AAS)**.

Although steroids are derivatives of a male sex hormone they may be taken by both men and women but use is generally much higher among males. People who use steroids generally fall into four categories:

- Sports people—steroids promote muscle growth and strength, reduce recovery time after training and enable intensive training for longer periods. The use of steroids is banned in almost all sports.
- Body builders—to increase muscle growth.
- People who work in the security industry, like bodyguards and bouncers—to increase body size and strength.
- Adolescents—often young males, who wish to copy the physical stature of sporting heroes.

As with any drug, there are risks associated with taking steroids. People may become dependent on the drug or may suffer withdrawal symptoms if they stop using it. Steroids can also have serious effects on a person's health depending on age, sex, level of exercise, the type of drug, how much is used and the purity of the drug. Risks are especially high for young people who are still growing. Table 22.3 shows some of the health risks associated with consumption of steroids.

Table 22.3 Possible health effects of steroid use

Males	Females	Both males and females
Baldness	Enlargement of the clitoris	Acne
Decreased production of testosterone	Irregularities of the menstrual cycle	Decreased immunity to disease
Development of breasts (gynaecomastia)	Permanent deepening of the voice	Headaches
Impotence	Shrinking breasts	High blood pressure
Infertility or decreased sperm production	Growth of body and facial hair	Increased muscle size and strength
Pain when urinating		Liver damage/jaundice
Prostate problems		Psychological changes such as mood swings, depression, aggression
Shrinking testes		Sleeplessness
		Water retention/bloating
		Increased cholesterol level

EXTENSION

There are an increasing number of articles and advertisements in the media concerning health issues for males. Using a variety of search techniques
Find out:

- what the major health issues facing men are today
- how health professionals are trying to encourage men to take more interest in their health
- what some of the consequences for society are if men as a group fail to take a greater interest in their health.

Working scientifically

Activity 22.1 Debating the issues

Human intervention in reproduction always provokes controversy. Some of the ethical issues associated with such intervention are mentioned in this chapter. People have a diversity of views on such issues and many of the issues make good topics for classroom debate or discussion. Ideas for topics are presented below. In cooperation with your teacher you could discuss or debate one or more of these or other issues.

Actively participate in any debate or discussion by keeping a record of the arguments for and against the topic. This could be done as a table with one column for arguments for and one for arguments against.

During the exchange of views on any issue remember to respect the opinions of others even though you may disagree with them.

(a) In-vitro fertilisation—good value or a waste of money?

'Money spent on in-vitro fertilisation techniques for a few people would be much better spent on medical research or on treatments that would benefit large numbers of people.' (Extract from a letter to an Australian newspaper)

Debate/discuss the pros and cons of in-vitro fertilisation.

(b) Abortion—should women have the right to choose?

'Abortion is the same as murder; it is the deliberate termination of a human life.' (Spokesperson for the Right to Life Association)

'Every woman has the right to decide on matters affecting her own body.' (Spokesperson for a pro abortion-on-demand lobby group)

Debate/discuss the issue that abortion should be available to any woman on demand.

(c) Cloning—playing God or helping humankind?

'Too many bits of the technology needed to produce a human clone are already being developed for other, widely accepted medical treatments or for research purposes. And because human cloning cannot go on looking difficult for long, . . . the doctors and scientists demanding a worldwide ban on cloning won't be able to delay someone, somewhere, making a success of it.' (Editorial, *New Scientist*, 17 January 1998)

Debate/discuss the arguments for and against cloning humans.

(d) Sex selection—should parents be able to balance their family so that not all children are of the same sex?

'*I'm dubious about its use for social purposes, not because I have a strong view that it's immoral, but because I think it's probably unwise to use a very heavy technology. Pre-implantation genetic diagnosis is a very involved treatment which involves invading an embryo, which involves IVF treatment and it seems to me to do that for a relatively trivial reason, like family balancing, is probably unwise.*' (Lord Robert Winston, IVF specialist, *Lateline*, 2004, television program, ABC Television, 30 August)

Debate/discuss whether sex selection should be available to any couple who wish to use it.

Activity 22.2 Formula milk products

Visit a supermarket or pharmacy and record the ingredients on a number of baby milk-formula products. Take note of the main ingredients.

1. How do they differ from the nutrients present in human milk? (Refer to Table 22.1.)
2. Would these differences have any significance for the nutrition of an infant?
3. What nutritional advice is given to mothers on the labels of formula milk products?

   Compare your observations with those of other members of the class so that a wide range of products can be compared.
4. In class discussion, decide whether the labelling of formula milk products is adequate. What changes would your class recommend?

REVIEW QUESTIONS

1. **(a)** What is sex selection?
   (b) Describe the two pre-implantation methods of sex selection.
   (c) How could sex be selected after implantation?
2. What is the cause of a genetic disease?
3. Explain the difference between a single gene disorder and a polygenic disease. Give an example of each.
4. Using an example explain what causes a person to have a chromosomal disorder.
5. What are the two main ways of preventing genetic disease?
6. Explain the difference between replacement therapy and gene therapy in the treatment of genetic diseases.
7. In the treatment of genetic disease, germ line therapy raises ethical questions that are not associated with somatic cell gene therapy. Explain the reasons for this.
8. Why are viruses ideally suited for use to transfer genes into cells?
9. List five advantages of breastfeeding.

10. **(a)** What is herd immunity?
(b) Why is herd immunity important in a population?

11. Anabolic steroids are legitimately prescribed for many illnesses. Why do people take anabolic steroids illegally?

APPLY YOUR KNOWLEDGE

1. Suppose that sex selection became freely available at very low cost. What would be some of the advantages and disadvantages to society if all couples were able to choose the sex of their children?

2. Explain why the results of the Human Genome Project will be very important in developing therapies for genetic diseases.

3. Table 22.1 compares the composition of colostrum, human milk and cow's milk. Examine the table and list the problems that could arise if a baby was fed on cow's milk instead of human milk.

4. Outline some of the factors that may influence a woman in reaching a decision on whether to breastfeed or bottlefeed her baby.

5. In 1998 a scientist claimed to have found a link between MMR (measles, mumps, rubella) vaccinations and the development of autism in children. The investigation was conducted on 12 children who had been given MMR vaccinations. There was no control group that did not receive the vaccine.
(a) How reliable do you think the results of such an investigation would be? Comment particularly on the number of children in the trial and the lack of a control.
(b) As a result of the publicity given to the 1998 claim of a link between MMR vaccination and autism many parents decided not to have their children vaccinated with MMR. What are the implications of such a decision for (i) the child, and (ii) society in general?

6. A 1999 survey of West Australian school students aged from 12 to 17 found that 4% of them had used steroids without a doctor's prescription. Suggest some reasons why young people may wish to take steroids.