

# 1

## Key principles of pathophysiology

Figure 1.1 Mendelian trait

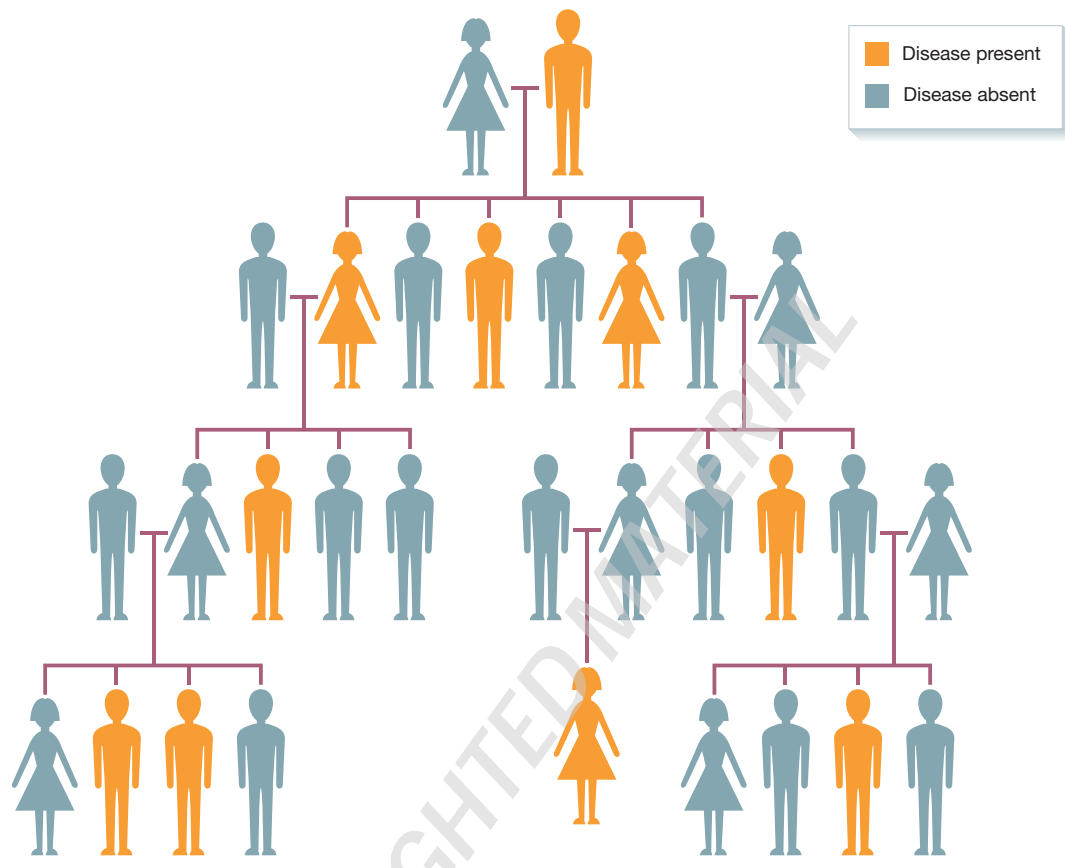
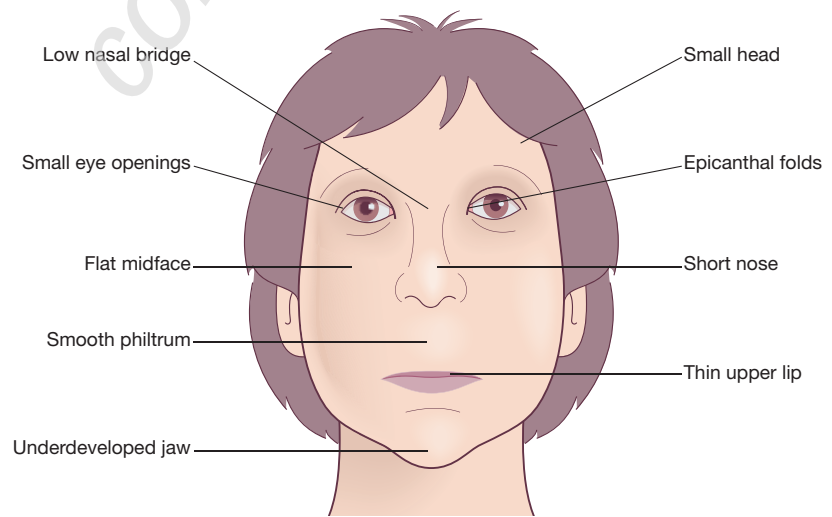


Figure 1.2 Foetal alcohol syndrome



## Pathophysiology versus pathology

While both terms indicate the study of disease, the term pathology is a broader term dealing with all aspects of a disease. This study is valuable for a physician or a pathologist who is also interested in the macro and microscopic characteristics of tissues and organs. On the other hand, in pathophysiology the focus is on the abnormal function of diseased organs, with application to diagnostic procedures leading patient care. Healthcare professionals are more concerned with pathophysiology when dealing with patients.

## Disease and aetiology

The study of the cause of disease is called aetiology. Aetiology is the preferred spelling in some countries, including the UK, whereas 'etiology' without an 'a' is used in the USA. The word 'aetiology' comes from the Greek *aitia*, cause + *logos*, discourse. Diseases are described as genetic, congenital or acquired.

### Genetic

In a disease where the cause is genetic, the person may have a defective gene that causes the disease. These defective genes are often passed on to children by parents. These abnormalities can range from a small mutation in a single gene to the addition or subtraction of an entire chromosome or set of chromosomes.

Some genetic diseases are called Mendelian disorders (Figure 1.1); they are caused by mutations that occur in the DNA sequence of a single gene. These are usually rare diseases; some examples are Huntington's disease and cystic fibrosis. Many genetic diseases are multifactorial – they are caused by mutations in several genes, compounded by environmental factors. Some examples of these are heart disease, cancer and diabetes.

### Congenital

In congenital disease, the genetic information is intact; however, problems with the intrauterine environment may result in congenital disorder. For example, cystic fibrosis is a genetic disorder, whereas foetal alcohol syndrome results from the mother's alcohol intake during pregnancy. This results in congenital abnormalities in a child who is genetically normal (Figure 1.2).

### Acquired

In this type of disease, the person develops the disease after birth as a result of direct or indirect contact with another person or the environment. Examples include tuberculosis, emphysema, chicken pox or acquired heart diseases.

## Signs and symptoms

A symptom is generally subjective, while a sign is objective. Any objective evidence of a disease, such as blood in the stool or a skin rash, is a sign – it can be recognized by the doctor, nurse, family members and the patient. However, stomachache, lower-back pain, fatigue, for example, can only be detected or sensed by the patient – others only know about it if the patient tells them. For example, pain can either be acute or chronic. An example of acute pain is abdominal pain, which is sudden and may last only a few

hours or longer. Common chronic pain complaints include headache, low back pain, cancer pain, arthritis pain, neurogenic pain (pain resulting from damage to the peripheral nerves or to the central nervous system itself), psychogenic pain (pain not due to past disease or injury or any visible sign of damage inside or outside the nervous system).

## Pathogenesis

In assessing a patient's signs and symptoms, conclusions can often be drawn about the pattern and development of a disease, in other words its pathogenesis. A typical pathogenesis involves kinds of tissue damage which produces certain effects. The progress of the disease can produce signs and symptoms throughout the course of the disease.

Another aspect of pathogenesis is the time over which the disease develops. Some may be acute, while others are chronic. Acute conditions have a rapid onset with short duration, while chronic conditions last for a longer period which could be from months to years.

## Investigations and diagnosis

In order to make a diagnosis, it may be necessary to carry out some investigations to confirm the diagnosis. Some of the investigations may be invasive, while others are not invasive. These may include blood test, CT scans, chest X-rays, endoscopy and many more.

Diagnosis is identification of a condition, disease, disorder or problem by systematic analysis of the background or history, examination of the signs or symptoms, evaluation of the research or test results, and investigation of the assumed or probable causes. It is from the diagnosis that care or treatment is prescribed.

## Treatment

Once a diagnosis is confirmed then the treatment can proceed. The treatment is either medical or nursing treatment. The aim of the treatment of a disease is to achieve a cure or minimize the patient's signs and symptoms to a degree where the patient can function near normality.

## Prognosis

Prognosis is a prediction of the chance of recovery or survival from a disease. Most doctors give a prognosis based on statistics of how a disease acts in studies on the general population. Prognosis can vary depending on several factors, such as the stage of disease at diagnosis, type of disease and even gender for example cancer.

Many factors can influence the prognosis of a patient with cancer. Among the most important are the type and location of the cancer, the stage of the disease (the extent to which the cancer has spread in the body) and how quickly the cancer is likely to grow and spread. Other factors that affect prognosis include the biological and genetic properties of the cancer cells (biomarkers), the patient's age and overall general health, and the extent to which the patient's cancer responds to treatment.