1 Key principles of pathophysiology

Figure 1.1 Mendelian trait.

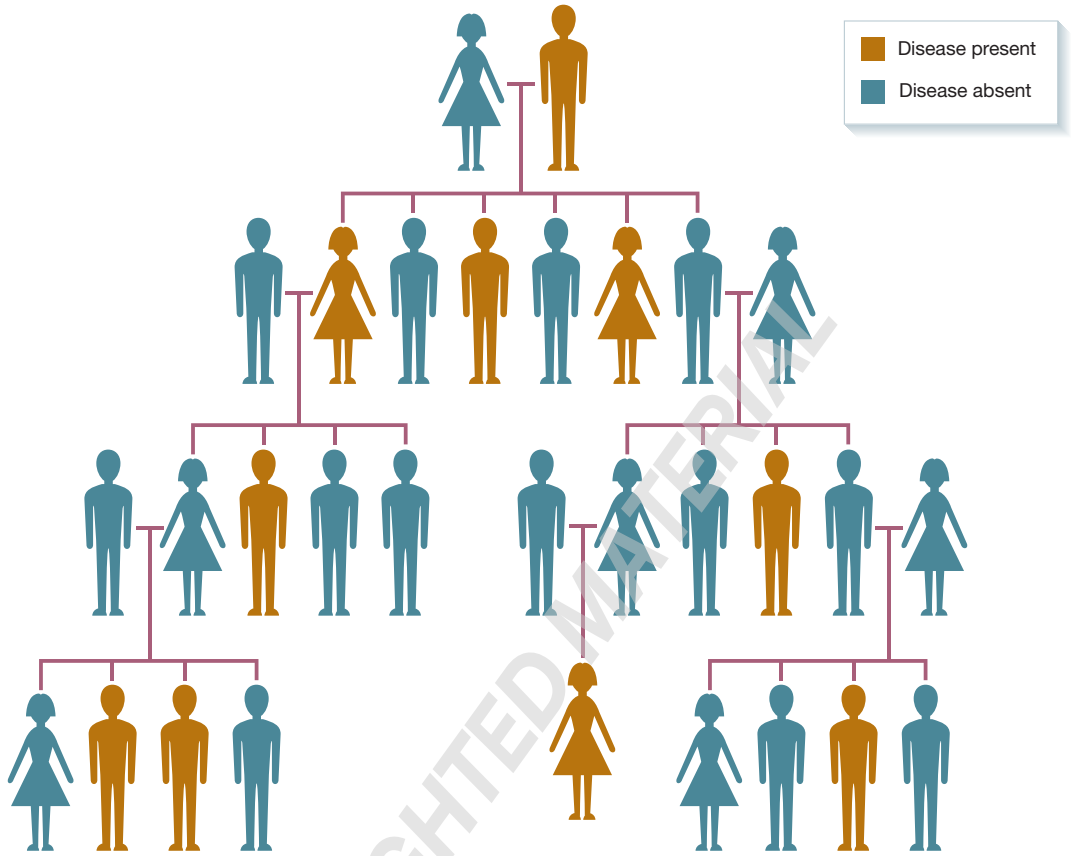
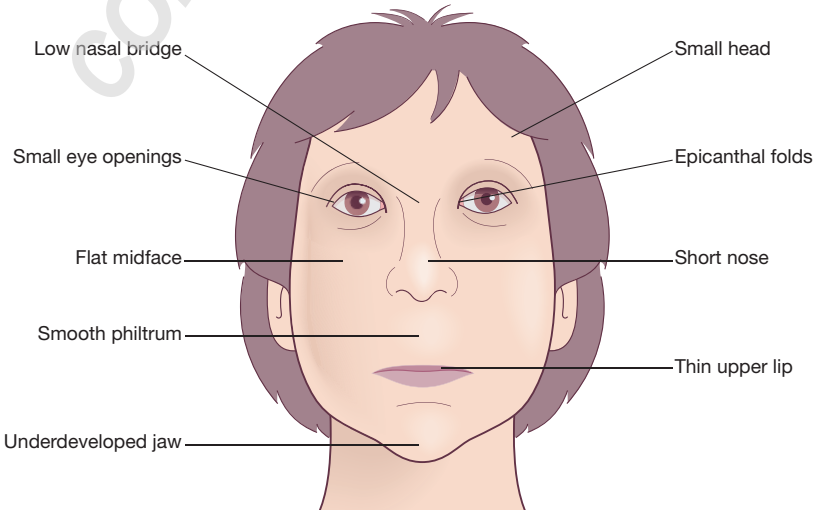


Figure 1.2 Fetal alcohol syndrome.



Pathophysiology versus pathology

Both of these terms indicate the study of disease. 'Pathology' is a broader term that deals with all aspects of a disease. This approach to disease is valuable for a physician or a pathologist who is 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 that lead to patient care. Healthcare professionals with face-to-face contact with patients are usually more concerned with pathophysiology when caring for patients.

Disease and aetiology

The study of the cause of disease is called aetiology, a fundamental concept in healthcare. Understanding aetiology enables healthcare professionals to identify why diseases occur, which factors contribute to their development and how these causes can be prevented or managed. Aetiology encompasses a wide range of influences, including biological factors (such as bacteria, viruses and genetic mutations), environmental factors (such as exposure to toxins or radiation) and lifestyle factors (such as diet, physical activity and smoking). By exploring the origins and mechanisms of disease, healthcare practitioners can make informed decisions about diagnosis, treatment and public health interventions.

The word 'aetiology' comes from the Greek *aitia*, cause + *logos*, discourse. Diseases are described as genetic, congenital or acquired.

Genetic

In diseases with a genetic origin, an individual may inherit a defective gene leading to the development of the condition. These faulty genes are often transmitted from parents to their children. Genetic abnormalities can vary widely, from a small mutation in a single gene to the loss or gain of an entire chromosome or set of chromosomes.

Some genetic conditions are known as Mendelian disorders (Figure 1.1), resulting from mutations in the DNA sequence of a single gene. These disorders include Huntington's disease and cystic fibrosis. Many genetic diseases, however, are multifactorial, meaning they arise from mutations in multiple genes combined with environmental influences. Common examples include 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 fetal alcohol syndrome results from the mother's alcohol intake during pregnancy. This results in congenital abnormalities in a genetically normal child (Figure 1.2).

Acquired

In this type of disease, the person develops the condition after birth as a result of direct or indirect contact with another person or the environment. Examples include tuberculosis, emphysema, chickenpox 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 recognised by the doctor, nurse, family members and the patient. However, stomach ache, lower-back pain and fatigue, for example, can only be detected or sensed by the patient – others only know about it if the patient tells them. Pain, for example, 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) and psychogenic pain (pain not due to past disease or injury or any visible sign of damage inside or outside the nervous system).

Pathogenesis

When assessing a patient's signs and symptoms, this can often identify the pattern and progression of a disease, which is known as its pathogenesis. Pathogenesis describes how a disease develops and causes tissue damage, leading to the observable clinical effects. As the disease progresses, it produces characteristic signs and symptoms that can change throughout its course, providing important clues for diagnosis and management.

Another key aspect of pathogenesis is the timeframe of disease development. Some diseases are acute, whereas others are chronic and develop more gradually, persisting over an extended period, often months or years. Understanding whether a condition is acute or chronic helps in planning appropriate investigations, treatment and follow-up care.

Investigations and diagnosis

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

Diagnosis is the 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, treatment can proceed (e.g. medical or surgical). The aim of the treatment of a disease is to achieve a cure or minimise the patient's signs and symptoms to a degree where the patient can function near normal.

Prognosis

Prognosis is a prediction of the chance of recovery or survival from a disease. Most prognoses given are 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, the type of disease and even gender.

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), and the patient's overall general health and the extent to which the cancer responds to treatment.

Clinical considerations

Linking the underlying disease mechanisms to a patient's signs and symptoms can identify the cause of illness, anticipate complications and select the most appropriate interventions. For instance, recognising that shortness of breath in heart failure results from fluid accumulation in the lungs helps guide treatment with diuretics and oxygen therapy. Similarly, understanding that pyrexia and inflammation are part of the body's immune response assists in distinguishing between infection and autoimmune disease. Applying pathophysiological principles in clinical practice enhances diagnostic accuracy, improves treatment outcomes and supports the delivery of safe, evidence-based and person-centred care.