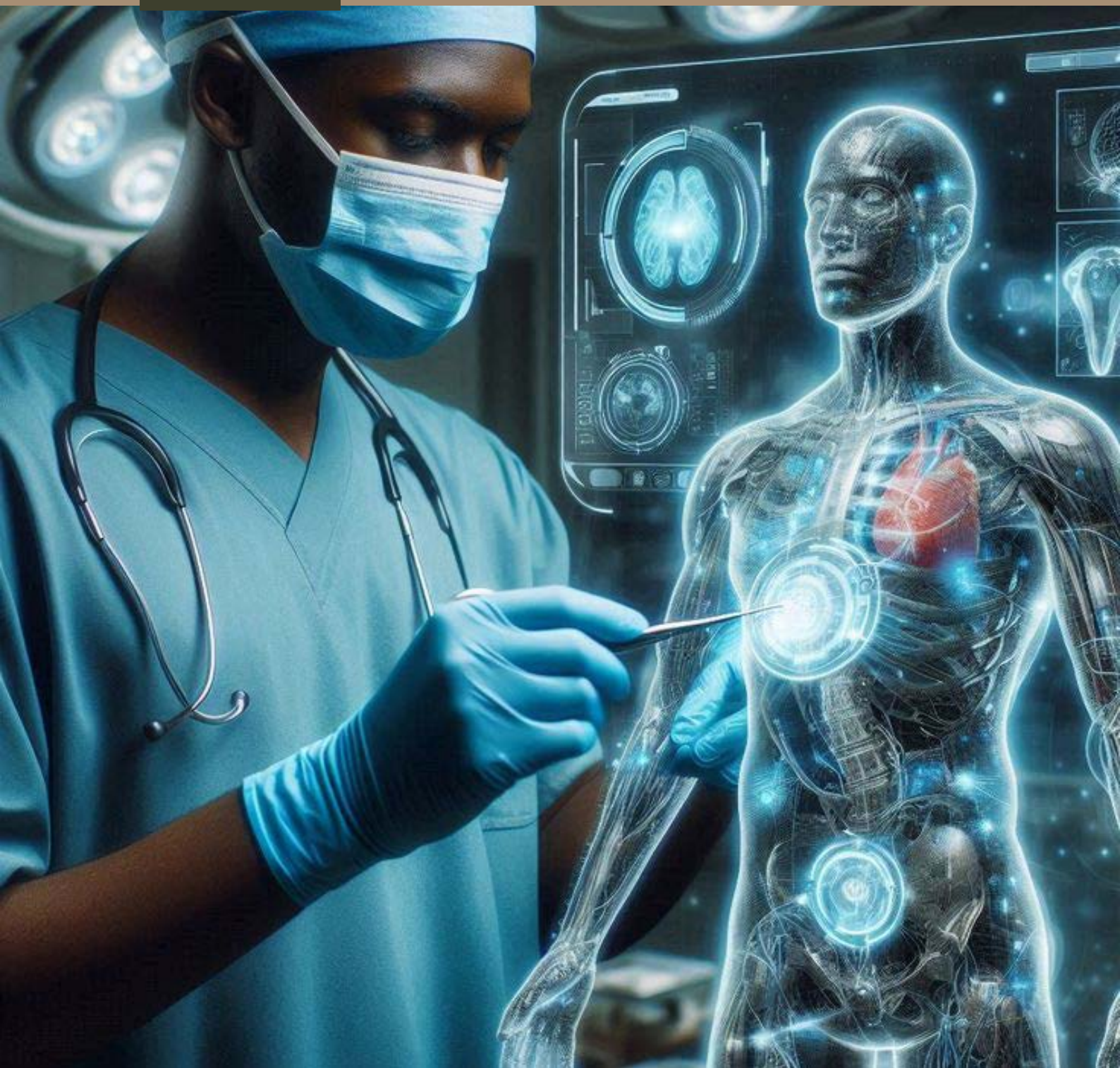
SECTION

# 4

# DISEASES AND DISORDERS



# Human Body Systems

## Diseases and Disorders

## INTRODUCTION

Welcome to this section. You will first explore diseases and disorders with specific examples. Understanding the differences between diseases and disorders is important for recognising how our bodies function and how they can be affected. A disease is a condition caused by microorganisms, such as bacteria or viruses. In contrast, a disorder is when there is a change from the normal way the body works or is structured. Both diseases and disorders show symptoms and signs, indicating that something is wrong with the body's normal processes. Our bodies have various ways to defend against infectious diseases, and we can also protect people through vaccination programs and public health measures. You will wrap it up with herd immunity. Make an effort to go through the activities and scenarios as well as make time for the extended reading materials.

### At the end of this section, you will be able to:

- Demonstrate an understanding of what a disease is and give examples.
- Explore how initial symptoms of an illness lead to diagnosis and treatment.
- Explore the symptoms and factors that contribute to sickle cell disease and how to curtail it in humans.

### Key Ideas

- Diseases can be caused by harmful microorganisms called pathogens. These include bacteria, viruses, fungi, and protozoa
- Diseases usually have specific symptoms and signs. They cause abnormal changes in the way our body looks or works
- A symptom is a change in your body or mind indicating that you might have a disease or health condition
- Symptoms can be different for each disease. They can be mild or very serious. Some diseases last for a short time (acute), while others last a long time or even for life (chronic)
- Diseases and disorders can affect any part of the body such as organs, tissues, cells, and sometimes the whole body.
- Symptoms of diseases are vital clues that aid healthcare providers in identifying, localising, evaluating and managing various health conditions.
- Symptoms also serve as the cornerstone of the diagnostic process, guiding further evaluation and treatment decisions to promote optimal patient outcomes and well-being.
- Symptoms provide valuable information to healthcare professionals, helping them identify the underlying condition and develop appropriate treatment plans.

- However, it is important to note that symptoms alone may not be enough for a definitive diagnosis and further medical tests and examinations may be necessary to confirm the cause of the symptoms.
- Sickle cell disease is a genetic disorder that affects red blood cells.
- The disease causes various symptoms, including pain, anaemia, and organ damage.
- Early detection, medical management, and lifestyle modifications can help manage sickle cell disease and improve quality of life.

# THE CONCEPT OF DISEASES AND DISORDERS: THE RELATIONSHIP BETWEEN THEM

## What is a Disease?

A disease can be defined as a pathological condition of an organism that impairs its normal physiological functioning. It is a condition caused by a microorganism.

### Key Facts About Diseases

1. Pathogens like bacteria, viruses, fungi, and protozoa can cause diseases.
2. Diseases are usually identified by specific symptoms, signs, and abnormal changes in the body's structure or function.
3. Disease symptoms can range from mild to severe and can be either short-term (acute) or long-lasting (chronic).
4. Diseases can affect any part of the body, including organs, tissues, cells, and the entire organism.

**Fun Facts: Did you know?**

**Some bacteria in our bodies are good for us! For example, gut bacteria help us digest food and produce vitamins.**

## What is a Disorder?

A disorder is a broad term sometimes used to describe any abnormal condition or deviation from the normal functioning or the structure of the body. It is a deviation from normal body function or structure.

### Key Facts About Disorders

1. Disorders can refer to physical, mental, or emotional disruptions that differ from normal patterns of health and well-being.
2. Disorders can appear in various forms and levels of severity, from mild to severe.

# Types of Diseases with Examples

Diseases can be grouped in different ways. Below are some common types based on causes and modes of transmission:

## 1. Infectious or Communicable Diseases

These diseases are caused by pathogens and can spread from person to person.

Examples include:

a. **Bacterial Infections**: Tuberculosis, Streptococcal Infections, Typhoid Fever, Pneumonia.

b. **Viral Infections**: Influenza (Flu), HIV/AIDS, COVID-19, Hepatitis.

c. **Fungal Infections**: Ringworm, Athlete's Foot, Candidiasis (yeast Infections).

d. **Vector-borne Parasitic Infections**: Malaria, Sleeping Sickness, Head Lice Infection, Elephantiasis, Bilharzia.

## 2. Non-infectious or Non-Communicable Diseases

These are conditions that are not caused by infections and cannot be passed from person to person. They often need long-term treatment.

Examples are given below:

a. **Cardiovascular Diseases**: Coronary Artery Disease (CAD), High Blood Pressure, Stroke.

b. **Cancer**: Breast Cancer, Lung Cancer, Prostate Cancer.

c. **Respiratory Diseases**: Asthma, Chronic Obstructive Pulmonary Disease (COPD).

d. **Musculoskeletal Diseases**: Osteoarthritis, Rheumatoid Arthritis.

# Types Of Disorders with Examples

Below are the various ways through which a deviation from normal body function or structure occurs and their specific examples.

1. **Genetic Disorders:** These are caused by changes in genes inherited from parents. Examples include

   a. **Sickle Cell Disease:** It is a genetic disorder where red blood cells are shaped like sickles, blocking blood flow and causing pain.

   b. **Haemophilia:** It is a genetic disorder where blood does not clot properly, causing too much bleeding even from small cuts or injuries.

   c. **Down Syndrome:** A genetic disorder caused by an extra chromosome, leading to developmental delays and physical traits.

d. **Cystic Fibrosis:** It is a genetic disorder that causes thick, sticky mucus to build up in the lungs and digestive system which leads to breathing problems and infections.

2. **Metabolic Disorders:** These disrupt normal metabolism, affecting how the body turns food into energy and removes waste. Below are examples:

   a. Diabetes (Type 1 and Type 2): A condition where the body cannot properly control blood sugar levels. Type 1 diabetes is when the body cannot produce insulin due to the destruction of insulin-producing cells in the pancreas, while Type 2 diabetes is when the body does not use insulin properly or does not produce enough insulin.

   b. Obesity: A condition where a person has too much body fat, which can lead to health problems.

3. **Neurological Disorders:** These affect the brain, spinal cord, and nerves, often due to structural or chemical abnormalities. Examples include:

   a. Autism: It is a developmental disorder that affects communication and behaviour, often making social interactions difficult.

   b. Cerebral Palsy: This is a group of disorders that affect movement and muscle tone, caused by damage to the brain before or at birth.

   c. Multiple Sclerosis: The immune system attacks the protective covering of nerves, causing communication problems between the brain and the rest of the body.

   d. Alzheimer's Disease: A brain disorder that causes memory loss and thinking problems over time.

   e. Parkinson's Disease: A nervous system disorder that causes tremors, stiffness, and movement difficulties.

   f. Epilepsy: A neurological disorder that causes recurring seizures due to abnormal brain activity.

4. **Mental Health Disorders:** These conditions affect mood, thinking, and behaviour. Examples are:

   a. Depression: It is a mental health disorder that causes persistent feelings of sadness and loss of interest in daily activities.

   b. Anxiety Disorders: These are mental health conditions characterised by excessive worry, fear, or nervousness

   c. Schizophrenia: It is a chronic and severe mental health disorder that affects how a person thinks, feels, and behaves

   d. Bipolar disorder: A mental health condition that causes extreme mood swings, including emotional highs (mania or hypomania) and lows (depression).

5. **Lifestyle-Related Disorders:** These are caused by unhealthy habits like poor diet, lack of exercise, and smoking. Examples are Type 2 diabetes, and hypertension (high blood pressure).

## Symptoms Associated with Some Diseases and Disorders

A symptom is a sign of a disease or disorder that a person feels or notices. Symptoms are usually reported by the person experiencing them and might not be visible to others. They can be physical, emotional, or cognitive (mental process), and they differ based on the condition.

### Below are Some Common Diseases and Their Associated Symptoms

1. **Influenza (Flu):** Symptoms include high fever, muscle aches, chills, fatigue, headache, sore throat, runny or stuffy nose, and cough.
2. **COVID-19:** Symptoms include fever, cough, shortness of breath, fatigue, muscle or body aches, loss of taste or smell, sore throat, congestion or runny nose, nausea or vomiting, and diarrhoea.
3. **Diabetes (Type 1 & 2):** Symptoms include increased thirst, frequent urination, fatigue, blurred vision, slow-healing wounds or sores, tingling or numbness in hands or feet, and unexplained weight loss.
4. **Malaria:** Symptoms include loss of appetite, hot and cold chills, fatigue or loss of energy, headaches, fever, nausea and vomiting, and anaemia (in severe cases).
5. **Asthma:** Symptoms include shortness of breath, wheezing (a high-pitched whistling sound during breathing), coughing (especially at night or early morning), and chest tightness or pain.
6. **Cardiovascular Disease for example before coronaryartery disease):** Symptoms include chest pain or discomfort (angina), shortness of breath, fatigue, rapid or irregular heartbeat, and dizziness or light-headedness.
7. **Common Cold:** Symptoms include runny or stuffy nose, sneezing, sore throat, cough, mild fatigue, and mild headache.

***Great! Now let us look at how the body responds to infections, cuts or injury***

## How the Human Body Responds to Infection

When the body encounters a harmful microorganism, it activates its immune system to fight against the infection. This response involves various mechanisms aimed at identifying, neutralising, and eliminating the invading pathogens.

1. The body's first line of defence includes the physical barriers like skin, stomach acid, mucus, and cilia in the lungs, as well as tears, to prevent pathogens from entering the body.

2. If a pathogen gets past these barriers, the immune system sends or deploys special cells such as macrophages and lymphocytes to find and kill them.
3. Macrophages engulf and digest (that is, they surround and eat up) pathogens and lymphocytes produce antibodies that bind to specific antigens on the pathogens, marking them for destruction. This process triggers an inflammatory response, characterised by redness, swelling, and heat, as the body works to eliminate the infection.
4. In the end, the immune response helps to heal the infection and makes the body better at fighting off the same pathogen in the future by establishing immunity.

   Lymphocytes and macrophages are cells of the immune system. They work together to identify and destroy pathogens. Lymphocytes include T-cells and B-cells.

   a. T-lymphocytes (T-cells) directly attack pathogens or help regulate the immune response.
   b. B-lymphocytes (B-cells) produce antibodies to fight specific pathogens.

**Can you guess how the immune system can distinguish the body cells from foreign particles?**

5. The immune system can distinguish between "self" cells and "non-self" cells **using markers** on their surfaces. This helps prevent the immune system from attacking the body's cells.
6. Though the immune system does well to recognize “self” cells from “non-self” cells sometimes the unexpected does happen and the immune system attacks the body’s own cells. This is called the **autoimmune response**. In autoimmune responses, the immune system mistakenly attacks the body's cells, leading to diseases like rheumatoid arthritis and lupus.

**Did you know?**

**Our immune system has a "memory" and can remember past infections to fight them off more quickly in the future.**

Let us now explore how the vaccination of the majority group can help save others.

## Herd or Community Immunity Against Transferable Diseases

When many people in a community become immune to a disease, it's called herd immunity or community immunity. This happens either because they got vaccinated or because they had the disease before. When enough people are immune, the disease cannot spread easily, which helps protect those who cannot be vaccinated, like babies, older people or people with weak immune systems.

## Here are the Ways Through Which Herd/Community Immunity is Achieved

Herd immunity is achieved mainly through vaccination, which helps the body build protection without getting sick, and by having a memory response that fights off the disease if it tries to come back.

a. **Vaccination**: When many people get vaccinated against a disease, their bodies develop antibodies that protect them from getting sick if they are exposed to the same pathogen in the future. Vaccines not only protect the person who gets vaccinated but also help protect the whole community by preventing the spread of infectious diseases. This helps to stop the spread of diseases like polio and smallpox.

b. **Immune Memory Response**: After recovering from an infection, the body remembers how to fight off the same germ if it comes back. This makes it hard for the germ to infect others and slows down its spread.

Let us go through the following activities to build more understanding of what we have discussed so far. Remember to attempt the review questions as well.

**Did you know?**

**The common cold can be caused by more than 200 different viruses?!**

## Activity 4.1: Symptoms of Flu

Scan the QR code 1 below or click here to watch a video on Symptoms of Flu (influenza).

From the video,

a. How do you know you have flu? (List the symptoms mentioned in the video)
b. Which ones have you seen in a relative or a friend?
c. Which people are at high risk of contracting flu?
d. List some of the measures a person suffering from flu can take

**Symptoms of Influenza (Flu)**

## Activity 4.2: List of diseases and disorders

From the following list of diseases and disorders: **Flu, asthma, COVID-19, Type 1 & 2 diabetes, stroke, depression, malaria, tuberculosis, sickle cell disease, candidiasis.**

Provide the following information

1. Type of disease or disorder (that is, infectious, non-communicable or disorder).

**Guide**

| Infectious diseases | Non-infectious diseases | Disorders |
|---|---|---|
| 1 | 2 | 3 |
| 4 | 5 | 6 |

   a. Can the condition spread from person to person?

      i. If yes, put it under infectious diseases.
      ii. If no, put it under non-communicable
      iii. If it is a deviation from normal body functioning or structure, then take it to disorders.

2. Causative agent of each disease or risk factor for disorders

   **Guide**

   a. Is the condition caused by a pathogen?
   b. If yes, specify if it is bacteria, fungi, viruses, vector-borne parasitic infection
   c. If no, specify the risk factors for those non-infectious disorders.
   d. State the specific pathogen or risk factors for non-communicable ones.

3. Symptoms of the disease and disorders.

   **Guide**

   a. Consider the signs you see when infected (List some of these signs or risk factors).
   b. Surf the internet, ask questions, or refer to textbooks if you are unsure about the causative agent and symptoms.

Well done! Now create posters of the diseases and disorders with their causative agents or risk factors and symptoms. Educate someone on how to prevent them. Let's save the world!

## Activity 4.3: Identifying 10 common diseases and disorders in Ghana and Africa

1. List those you have had before. What symptoms did you have?
2. List those you have seen in a friend or colleague. Identify the symptoms noted
3. What disorders are you familiar with?
4. Surf the internet or make enquiries for more information on each
5. Make a poster about each with their causes and symptoms.

Educate as many people as you can on these diseases and disorders and how to avoid them

**Self-Reflection**

1. How many of the diseases or disorders were you able to provide the required information?
2. Have I been able to apply the knowledge acquired in this stage to solve real-life scenarios?
3. What were the challenges you encountered in solving the scenarios?
4. How can I use the knowledge acquired here to explain the concept of diseases and disorders to my colleagues?
5. Do I need to review the content again for vital information I could not grab well?

## Extended Reading

- To learn more about the Immune System, how it works and its role in protecting the body, (https://www.nature.com/articles/s41577-020-0391-5)
- The Role of Macrophages During Mammalian Tissue Remodelling and Regeneration Under Infectious and Non-Infectious Conditions. (https://www.frontiersin.org/journals/immunology/articles/10.3389/fimmu.2021.707856/full)
- To read more on herd immunity, use (https://www.pnas.org/doi/full/10.1073/pnas.2107692118)

# THE IMPORTANCE OF THE SYMPTOMS OF DISEASES TO DIAGNOSIS.

Welcome to today's lesson! Let us first find the meaning of diagnosis. Diagnosis refers to the process of identifying a disease or disorder based on the evaluation of symptoms, signs, medical history, physical examinations, and diagnostic test results. A proper diagnosis is important for understanding the underlying cause of a patient's condition and determining the most appropriate course of action.

Diagnosis and treatment of illnesses (diseases and disorders) based on initial symptoms are crucial aspects of medicine. Physicians use a systematic approach (scientific method) to assess a patient's symptoms, medical history, physical examination, and sometimes additional diagnostic tests to arrive at a diagnosis and recommend appropriate treatments.

Here, you will explore how initial symptoms of illnesses can be pivotal in diagnosis and treatment.

## Do You Recall the Meaning of Symptoms?

Symptoms are how the body tells us something is wrong and are the first signs that make people see a health worker.

They help health workers figure out what might be causing the problem. By paying close attention to symptoms, health workers can find out what's wrong more quickly and start the right treatment.

Knowing and understanding symptoms is key to diagnosing diseases early and getting better results from treatments.

### Below are Some Ways of Using Symptoms for Healthcare Delivery

1. **Diagnosis:** Symptoms are the main signs that help health workers think about what illnesses or conditions might be affecting a patient. By paying close attention to what a patient says about their symptoms and doing a detailed check-up, health workers can narrow down the possible diagnoses and find the most likely cause of the patient's health problem
2. **Treatment Planning:** Symptoms give important information for creating a good treatment plan. Different diseases and conditions need specific treatments, medicines, or therapies. By understanding the symptoms, health workers can design treatments that target the cause of the illness. For example, if a patient has a cough, fever, and difficulty breathing, these symptoms might suggest pneumonia, and the health worker can prescribe antibiotics to treat the infection.
3. **Monitoring Progress:** After starting treatment, health workers keep track of the patient's symptoms to see how they respond to the treatment and make changes if needed. Improvement or worsening of symptoms helps them understand how well the treatment is working.
4. **Early Detection:** Some diseases start with mild or non-specific symptoms. Health workers who notice these early signs can quickly diagnose and treat the illness. This can lead to better results and prevent the illness from getting worse.
5. **Public Health Surveillance:** Identifying and watching specific symptoms in a population is important for public health. It helps health authorities find disease outbreaks or trends, take proper public health actions, and use resources effectively. During the COVID-19 pandemic, tracking symptoms like runny nose, cough, and loss of taste or smell helped authorities detect and manage outbreaks, implement lockdowns, and allocate medical supplies where they were needed most.

## How Diagnosis and Treatments are Deduced from Initial Symptoms

1. **Patient History:** Health workers begin by collecting a detailed history of the patient. This includes information about past medical issues, family health history, lifestyle, and any current medications. Understanding the patient's history helps in identifying patterns or risk factors that might be relevant to the current symptoms.
2. **Major Complaint:** The next step is to identify the major complaint, which is the main reason the patient sought medical help. This primary symptom or issue guides the initial focus of the diagnosis process. For example, if a patient comes in with chest pain, this symptom will be the primary focus initially.
3. **Open-ended Questions:** Health workers use open-ended questions to allow patients to describe their symptoms in their own words. This approach ensures that all relevant details are captured. For example, a doctor might ask, "Can you describe how you've been feeling recently?" to gather comprehensive information about the patient's condition.
4. **Review of Systems:** A review of systems involves asking about symptoms related to different body systems (e.g., cardiovascular, respiratory, gastrointestinal). This helps to identify any other issues that might be related to the major complaint. For example, a patient with chest pain might also be asked about shortness of breath, dizziness, or stomach pain.
5. **Physical Examination:** A thorough physical examination is conducted to check for signs that support or rule out possible diagnoses. This may include checking vital signs like temperature, pulse, and blood pressure, as well as examining specific areas of concern. For instance, in a patient with chest pain, the doctor might listen to the heart and lungs and check for signs of heart failure or lung issues.
6. **Additional Diagnostic Tests:** Based on the initial findings, health workers may order additional tests to gather more information. These could include blood tests, imaging studies (like X-rays or MRIs), or specialized tests like biopsies. For example, a chest X-ray might be ordered for a patient with chest pain to check for pneumonia or other lung conditions.
7. **Refining the Diagnosis:** With the information from the patient history, physical examination, and diagnostic tests, health workers refine the diagnosis. They narrow down the list of possible conditions to identify the most likely cause of the symptoms. This process often involves ruling out less likely conditions and focusing on the most likely ones.
8. **Treatment Decision:** Once a diagnosis is made, a treatment plan is created. The plan is tailored to address the underlying cause of the symptoms. For instance, if the diagnosis is a bacterial infection, the treatment might include antibiotics. If the diagnosis is a heart attack, the treatment might involve medications, lifestyle changes, and possibly surgical interventions.
9. **Monitoring and Adjusting:** After treatment begins, health workers monitor the patient's progress to see how well they are responding. This involves regular check-ups and possibly additional tests. If the patient's symptoms improve, it indicates that the treatment is effective. If the symptoms worsen or new symptoms appear,

the treatment plan may need to be adjusted. For example, if a patient with pneumonia does not respond to initial antibiotics, a different antibiotic or additional treatments might be needed.

## Activity 4.4: Symptoms and Diagnosis

Scan the QR codes below to watch videos on how symptoms aid in the diagnosis and treatment of diseases.

**Video 1**

**Video 2**

## Activity 4.5: Understanding How Symptoms Lead to Diagnosis and Treatment

**Objective:** This activity will help you learn how initial symptoms can lead to the diagnosis and treatment of illnesses.

**Instructions:** You will perform a role play where you act out how initial symptoms lead to the diagnosis and treatment of an illness, like malaria or the flu. Form groups of three students and each student should choose one of the following roles: Doctor, Nurse, or Patient.

**Roles and Responsibilities:**

1. **Doctor:**
   a. Ask the patient about their symptoms.
   b. Use the information to figure out what illness the patient might have.
   c. Decide on the treatment and explain it to the patient.
2. **Patient:**
   a. Pretend to have symptoms of an illness (like fever, headache, or cough).
   b. Answer the doctor's questions honestly and clearly.
3. **Nurse/Assistant:**
   a. Help both the doctor and the patient.
   b. Take notes on what the patient says and what the doctor does.

**Steps:**

1. **Prepare:** Decide who will be the doctor, nurse, and patient. Think about the symptoms and what the illness might be.
2. **Role Play:**
   a. Patient: describe your symptoms to the doctor.
   b. Doctor: ask questions to understand more about the symptoms.
   c. Nurse: take notes and help as needed.
   d. Doctor: explain the diagnosis and treatment to the patient.

3. **Discussion:** After the role-play, discuss what happened. Did the doctor ask good questions? Did the patient give clear answers? How did the nurse help?

   **Example Scenario:**

   ***Illness: Malaria***

   - **Patient:** Complains of fever, chills, headache, and feeling very tired.
   - **Doctor:** Asks when the symptoms started, if the patient has travelled recently, and checks for signs like a high temperature.
   - **Nurse:** Writes down the patient's symptoms and the doctor's questions and findings.

**Self-Reflection**

Reflect on a time when you or someone you know experienced an illness. What were the initial symptoms, and how did recognising and reporting these symptoms affect the diagnosis and treatment received?

# GENETICS AND HEREDITY

All organisms receive their genetic makeup from their parents. This genetic makeup determines how the cells of an organism are built and function. Through heredity, specific characteristics or traits from parents are passed down to their offspring if these traits are determined by genes. One example of an inherited trait is sickle cell disease. In this module, you will delve into the symptoms, causes, and management strategies for sickle cell disease.

Let us get started!

## What is Sickle Cell Disease?

Sickle cell disease (SCD) also called Sickle Cell Anaemia, is a genetic disorder characterised by a distortion in the shape of the red blood cell. Haemoglobin is a protein in red blood cells responsible for carrying oxygen throughout the body. Normally, red blood cells are flexible and concave, allowing them to flow smoothly through blood vessels (See Fig. 4.1).

However, in individuals with sickle cell disease, the haemoglobin is sickle-shaped, causing red blood cells to become rigid and take on a crescent or "sickle" shape. These abnormal cells can get stuck in small blood vessels, leading to various complications (Fig. 4.1).

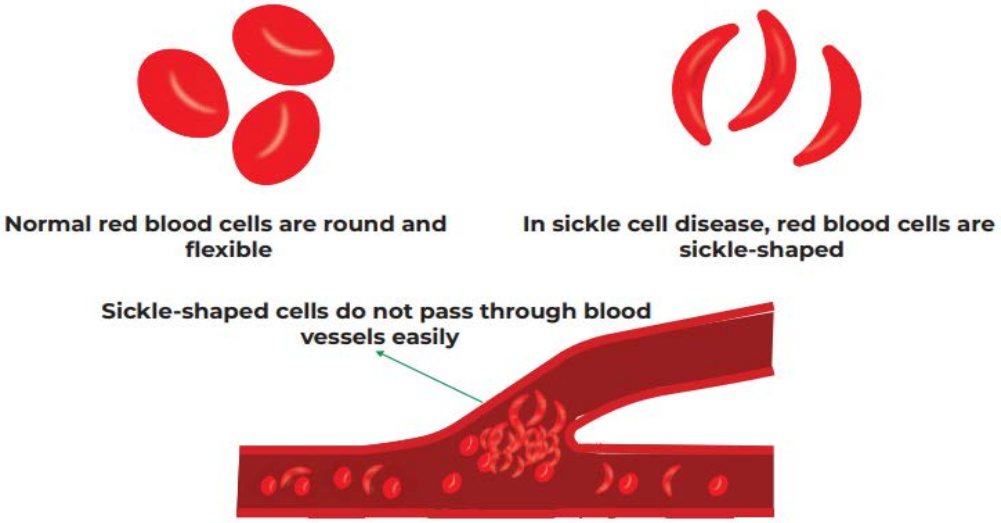



**Fig. 4.1:** Normal red blood cell and a sickle-shaped red blood cell

## Symptoms and Complications of Sickle Cell Disease

Sickle Cell Disease can cause many different symptoms, and how severe and how often these symptoms occur can vary for everyone.

### Below are Some Symptoms and Complications That Usually Occur

1. **Painful Crises:** pain crises are one of the hallmark symptoms of sickle cell disease. These episodes are characterised by severe and acute pain that can occur in various body parts, such as the chest, abdomen and joints. Pain crises may be triggered by factors like infection, dehydration, cold weather, stress, or high altitudes.
2. **Anaemia:** Sickle cells have a shorter lifespan than normal red blood cells, leading to chronic anaemia. Anaemia can cause fatigue, weakness, and paleness. Organ Damage: repeated sickling and blood vessel blockages can lead to the damage of various organs, including the spleen, liver, kidneys, and lungs. This can result in organ dysfunction and long-term complications.
3. **Fatigue:** SCD can lead to persistent fatigue and lack of energy. This is because of the reduction in the surface area of the red blood cell which makes it impossible for enough oxygen to be carried by the red blood cell.
4. **Swelling of Hands and Feet:** sickle cells can block blood flow in small blood vessels, causing swelling in the hands and feet, particularly in infants and young children.
5. **Frequent Infections:** sickle cell disease weakens the immune system, making individuals more susceptible to infections, particularly those caused by bacteria.
6. **Delayed Growth and Development:** Children with sickle cell disease may experience delayed growth and development compared to their peers.

7. **Jaundice:** Sickle cell disease can cause the breakdown of red blood cells, leading to an increased level of bilirubin in the blood. This can result in jaundice, which is characterised by yellowing of the skin and eyes.
8. **Organ Damage:** repeated sickling and blood vessel blockages can lead to the damaging of various organs, including the spleen, liver, kidneys and lungs. This can result in organ dysfunction and long-term complications.
9. **Acute Chest Syndrome: this** is a severe and potentially life-threatening complication of sickle cell disease, characterised by chest pain, fever, and difficulty in breathing. It is often caused by infection or blockage of blood vessels in the lungs.
10. **Stroke:** in children with sickle cell disease, there is an increased risk of stroke due to blood vessel blockage in the brain.

## How Sickle Genes are Transferred from Parents to Offspring Through Cross-Mapping

Cross mapping, also called Punnett square analysis, is a simple genetic tool used to predict the possible genotypes (gene combinations) and phenotypes (physical traits) of offspring when two parents with known genetic traits reproduce. For Sickle cell disease (SCD), it helps if we understand how sickle cell genes are passed from parents to children.

We have two genes for each characteristic, which can be dominant or recessive. The gene for SCD is recessive. This means that if you have only one SCD gene, it doesn't show because the normal dominant gene hides it, making you appear healthy. However, you are a carrier of the gene, which could be risky for your children.

To have SCD, you must inherit two recessive genes, one from each parent. If both parents carry the recessive gene there is a 25% chance of you getting the disease. This can be seen in Table 4.1.

In this situation, both parents are carriers of the SCD gene HbS. The normal gene is HbA.

**Table 4.1: Punnett square for Parent 1 (HbA/HbS) and Parent 2 (HbA/HbS)**

| Parents | HbA | HbS |
|---|---|---|
| **HbA** | **HbA/ HbA** | **HbA/HbS** |
| **HbS** | **HbA/HbS** | **HbS/HbS** |

# Review Questions

## Review Questions 4.1

**Scenario 1: Cut on the Skin**

You accidentally cut your finger while cooking. The cut is small but deep enough to draw blood.

1. Describe the body's first line of defence in response to the cut.
2. How does the body prevent pathogens from entering the body through the cut?
3. Explain how macrophages and lymphocytes would be involved in this scenario.
4. Discuss the importance of keeping the wound clean and protected during the healing process.

**Scenario 2: Respiratory Infection**

You inhaled airborne pathogens while walking through a crowded area, leading to a respiratory infection.

1. How did the pathogens enter the body in this scenario?
2. Describe the role of the mucous membranes in the respiratory tract as a first line of defence.
3. What happens if pathogens breach the mucous membranes and reach the lungs?

## Review Questions 4.2

1. Why is it important to recognise and report symptoms like fever and headache early? How can this help in getting the right treatment?
2. In cases where the initial symptoms are vague or non-specific, how do healthcare professionals navigate the diagnostic process to reach an accurate diagnosis and initiate appropriate treatment?
3. Imagine you are a health worker in a rural clinic. A 30-year-old woman comes in complaining of general fatigue, mild headaches, and occasional dizziness for the past two weeks. She hasn't noticed any other specific symptoms and says her health has otherwise been good.
    a. Describe the steps you would take to diagnose her condition given her vague and non-specific symptoms.
    b. What questions would you ask her to gather more information?
    c. What initial diagnostic tests might you consider and why?
    d. How would you use the information from her responses and test results to narrow down the possible diagnoses?

## Review Questions 4.3

1. A 10-year-old child shows the following symptoms, chronic pain, fatigue, jaundice, and susceptibility to infections. Identify the suspected condition of this child. Describe three complications of this disease.
2. How is sickle cell disease inherited, and what factors contribute to its development?
3. Describe strategies for managing and curbing sickle cell disease in human

## Self-Assessments

**Self-assessment 1:** ***Understanding Sickle Cell Disease.***

Reflect on the overview of sickle cell disease. Can you describe the symptoms and complications associated with the condition? Do you understand how sickle cell disease is inherited?

**Self-reflection 2:** ***Management Strategies.***

Think about the lifestyle modification activity. Were you able to identify effective strategies for managing sickle cell disease? How do these strategies contribute to improving the quality of life for individuals with the condition?

# Answers To Review Questions

## Answers to Review Questions 4.1

**Hint 1:**

1. The body's first line of defence includes physical barriers like the skin, which prevents pathogens from entering. So, when the skin is cut, it triggers an immediate response to prevent infection.
2. The formation of a blood clot, along with the release of antimicrobial substances by skin cells, helps to prevent pathogens from entering the body through the cut.
3. Macrophages arrive at the site to engulf and digest any pathogens that may have entered. Lymphocytes produce antibodies that target specific antigens on pathogens, marking them for destruction.
4. Keeping the wound clean and protected prevents further contamination and infection, allowing the body's natural healing processes to proceed effectively.

**Hint 2:**

1. Pathogens enter the body through inhalation of airborne particles, which can contain bacteria or viruses.
2. Mucous membranes trap pathogens in mucus, and cilia move the mucus out of the respiratory tract to prevent infection.
3. If pathogens reach the lungs, the immune system activates macrophages and lymphocytes to detect and destroy the pathogens.

## Answers to Review Questions 4.2

1. Recognising and reporting symptoms like fever and headache early is important because it helps health workers identify the illness quickly. This leads to timely and accurate treatment, which can prevent the illness from getting worse and promote faster recovery.
2. When initial symptoms are vague or non-specific, healthcare professionals use a systematic approach that includes taking a detailed patient history, asking thorough questions, conducting a comprehensive physical examination, and ordering initial diagnostic tests. This thorough and methodical process helps them gather the necessary information to reach an accurate diagnosis and initiate appropriate treatment.
3. Answers to this question differ from student to student.
    a. To diagnose her condition, I would start with a detailed patient history, asking about her daily activities, diet, sleep patterns, stress levels, and any recent changes in her life.
    b. Questions I would ask include: "When did you first start feeling these symptoms?", "Have you experienced any recent illness or stress?", "Are you

eating and sleeping well?", and "Have you noticed any other changes in your health, no matter how small?"

c. Initial diagnostic tests might include blood tests to check for anaemia, thyroid function, and blood sugar levels, as well as a basic metabolic panel to rule out any electrolyte imbalances.

d. Using the information from her responses and test results, I would look for patterns or abnormalities that could indicate conditions like anaemia, thyroid issues, or diabetes, helping me to narrow down the possible diagnoses and plan the appropriate treatment.

## Answers to Review Questions 4.3

1. The Symptoms listed indicate the child suffers from sickle cell disease. Three complications of sickle cell disease may include acute chest syndrome, stroke, and organ damage.
2. Sickle cell disease is inherited when both parents carry the sickle cell trait and pass it on to their child. The presence of an abnormal haemoglobin molecule (haemoglobin S) causes red blood cells to become rigid and sickle-shaped. Environmental factors such as dehydration and extreme temperatures can trigger sickle cell crises.
3. Management strategies for sickle cell disease include staying hydrated, avoiding triggers, seeking medical care when necessary, and genetic counselling for family planning.

## Hints to Self-Assessment 1: Understanding Sickle Cell Disease.

**Symptoms:** Sickle cell disease (SCD) can cause episodes of pain (known as sickle cell crises), fatigue, swelling in the hands and feet, frequent infections, delayed growth, and vision problems.

**Complications:** SCD can lead to severe complications such as acute chest syndrome, stroke, organ damage (especially to the spleen, liver, and kidneys), and increased risk of infection. It can also cause chronic pain, leg ulcers, and gallstones.

**Inheritance:**

Sickle cell disease is inherited from both carrier parents. This means a child must inherit two copies of the sickle cell gene (one from each parent) to have the disease. If both parents are carriers of the sickle cell trait (HbAS), there is a 25% chance with each pregnancy that the child will have SCD (HbSS), a 50% chance that the child will be a carrier (HbAS), and a 25% chance that the child will inherit normal haemoglobin (HbAA).

## Hint to Self-Assessment 2: Management Strategies

**Effective Strategies:**

- **Hydration:** Drinking plenty of water helps prevent dehydration, which can trigger sickle cell crises.

- **Pain Management:** Using medications, such as over-the-counter pain relievers or prescription opioids, and other techniques like warm baths and physical therapy can help manage pain.
- **Regular Check-ups:** Frequent medical check-ups with a healthcare provider can monitor and manage complications.
- **Healthy Lifestyle:** Eating a balanced diet, avoiding extreme temperatures, and avoiding high altitudes help manage symptoms and prevent complications.
- **Hydroxyurea:** This medication can reduce the frequency of pain episodes and the need for blood transfusions.

**Contribution to Quality of Life:**

These strategies help minimize pain and reduce the frequency of sickle cell crises, which can significantly improve daily functioning and overall well-being. Regular medical care and preventive measures reduce the risk of severe complications, while medications like hydroxyurea can enhance the quality of life by decreasing the severity of symptoms. Overall, effective management strategies enable individuals with SCD to lead healthier and more active lives.

## Extended Reading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# Acknowledgments




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