Epilepsy

Epilepsy, a condition/neurological disorder that causes repeated bursts of electrical activity in the brain. Epilepsy is the finite number of episode while seizure is multiple numbers of episodes.

Types of epilepsy

1. Generalised seizure- Tonic-clonic, Absence
2. Partial seizure- Simple partial Seizure, Complex partial seizure

Generalised Seizure

This type of seizure affects the entire body.

- **Tonic-Clonic (Grandmal epilepsy):** Aura (Light dazzle in front of eyes) — Muscle contraction — micturition, salivation — Respiration stops for 2 Sec — Followed by violent jerk, Sleep (0-2 min).
- **Absence seizures:** Absence seizures, previously known as petit mal seizures, often occur in children and are characterized by absence in body movements such as eye blinking or lip smacking. These seizures may occur in clusters and cause a brief loss of awareness.

Partial seizure

This type of seizure does not affect the whole body rather they affect any particular part of body.

- **Simple partial Seizure:** This type of seizure originates from a specific part of brain and affects only a single part of body such as leg, hand.
- **Complex Partial Seizure:** This type of seizure originates from more than one part of brain, and affects more than one part of body such as legs and hand.

Cause

Epilepsy has no identifiable cause in about half the people with the condition. Some are outlined below:

- **Genetic influence:** Researchers have linked some types of epilepsy to specific genes, but for most people, genes are only part of the cause of epilepsy. Certain genes may make a person more sensitive to environmental conditions that trigger seizures.
- **Head trauma.** Head trauma as a result of a car accident or other traumatic injury can cause epilepsy.
- **Brain conditions.** Brain conditions that cause damage to the brain, such as brain tumors or strokes, can cause epilepsy. Stroke is a leading cause of epilepsy in adults older than age 35.
- **Infectious diseases.** Infectious diseases, such as meningitis, AIDS and viral encephalitis, can cause epilepsy.
- **Prenatal injury.** Before birth, babies are sensitive to brain damage that could be caused by several factors, such as an infection in the mother, poor nutrition or oxygen deficiencies. This brain damage can result in epilepsy or cerebral palsy.
- **Developmental disorders.** Epilepsy can sometimes be associated with developmental disorders, such as autism and neurofibromatosis.

Diagnosis

- **Blood tests:** Your doctor may take a blood sample to check for signs of infections, genetic conditions or other conditions that may be associated with seizures.
- **Electroencephalogram (EEG):** This is the most common test used to diagnose epilepsy.doctors attach electrodes to your scalp with a paste-like substance. The electrodes record the electrical activity of your brain.
- **Computerized tomography (CT) scan:** A CT scan uses X-rays to obtain cross-sectional images of your brain.
- **Magnetic resonance imaging (MRI):** An MRI uses powerful magnets and radio waves to create a detailed view of your brain.
- **Functional MRI (fMRI):** A functional MRI measures the changes in blood flow that occur when specific parts of your brain are working.
- **Positron emission tomography (PET):** PET scans use a small amount of low-dose radioactive material that's injected into a vein to help visualize active areas of the brain and detect abnormalities.
- **Neuropsychological tests:** In these tests, doctors assess your thinking, memory and speech skills. The test results help doctors determine which areas of your brain are affected.

Treatment

Medication

Phenytoin
Valproic Acid
Gabapentin

Surgery

When medications fail to provide adequate control over seizures, surgery may be an option. With epilepsy surgery, a surgeon removes the area of your brain that's causing seizures.

Pathophysiology

Due to several factors (head injury, infection) — Imbalance between excitatory neurotransmission and inhibitory neurotransmission takes place. This causes depolarisation of neurons occurs in very large numbers, thus normal impulse contraction is disturbed — Excessive discharge of electric current takes place from cortical and subcortical region of brain— These currents radiates throughout our body causing muscle contraction and seizure.

Parkinson's disease

Parkinson disease is a progressive neurodegenerative disorder characterised by rigidity, tremor and hypokinesia (Problem in movement) with secondary manifestation as:

- Tremor at rest
- Pin rolling movement
- Clog wheel movement
- Mask like face
- Sialorrhoea

Causes

- **Genetic mutation:** Researchers have identified specific genetic mutations that can cause Parkinson's disease.
- **Environmental factors:** Exposure to certain toxins or environmental factors may increase the risk of later Parkinson's disease
- **Lewy bodies: small particle sometimes get struck in the brain thus causing blockage.**
- **Age:** People usually develop the disease around age 60 or older.

Pathophysiology

Parkinson's disease is a degeneration of neuron in substantia nigra par compacta region and nigrostatum tract of brain. Neuron in these region produces dopamine in brain thus degeneration of neuron leads to the deficiency of dopamine, thus leading to the disturbance of normal muscle tone and its coordination.

Symptoms

Tremor: can occur at rest, in the hands, limbs
Muscular: stiff muscles, difficulty standing, difficulty walking, difficulty with bodily movements.
Sleep: daytime sleepiness, early awakening, nightmares, restless sleep, or sleep disturbances
Whole body: fatigue, dizziness, poor balance, or restlessness
Cognitive: amnesia, confusion in the evening hours, dementia, or difficulty thinking and understanding
Speech: difficulty speaking, soft speech, or voice box spasms
Mood: anxiety or apathy
Nasal: distorted sense of smell or loss of smell
Urinary: dribbling of urine or leaking of urine
Facial: jaw stiffness or reduced facial expression
Also common: blank stare, constipation, depression, difficulty swallowing, drooling, falling, fear of falling, loss in contrast sensitivity, neck tightness, small handwriting, trembling, unintentional writhing, or weight loss

Diagnosis

No specific test exists to diagnose Parkinson's disease.
Parkinson's disease based on your medical history
A review of your signs and symptoms
A neurological and physical examination.
Imaging tests — such as MRI, CT, ultrasound of the brain.

Treatment

All medication targets to increase the level of dopamine in brain by administering dopamine from outside or preventing the metabolism of dopamine which is produced in brain.

- Carbidopa
- Levodopa
- Dopamine agonists-Bromcriptine
- MAO B inhibitors- Selegiline
- Catechol O-methyltransferase (COMT) inhibitors- Tolcapone

Stroke

A stroke occurs when the blood supply to part of your brain is interrupted or reduced, depriving brain tissue of oxygen and nutrients. Within minutes, brain cells begin to die.

Causes

Ischemic stroke

Ischemic strokes occur when the arteries to your brain become narrowed or blocked, causing severely reduced blood flow (ischemia). It occurs when a blood clot (thrombus) forms in one of the arteries that supply blood to your brain.

Haemorrhagic stroke

Haemorrhagic stroke occurs when a blood vessel in your brain leaks or ruptures thus blood comes out. Brain haemorrhages can result from many conditions that affect your blood vessels. These include:

- Uncontrolled high blood pressure (hypertension)
- Overtreatment with anticoagulants (blood thinners)
- Weak spots in your blood vessel walls (aneurysms)

Symptoms

- Trouble with speaking and understanding.
- Paralysis or numbness of the face, arm or leg.
- Trouble with seeing in one or both eyes.
- Headache
- Trouble with walking
- Memory loss
- Difficulty talking or swallowing
- loss of muscle movement

Diagnosis

1. Physical examination
2. Blood tests- for excess of glucose level
3. Computerized tomography (CT) scan
4. Magnetic resonance imaging (MRI)
5. Echocardiogram

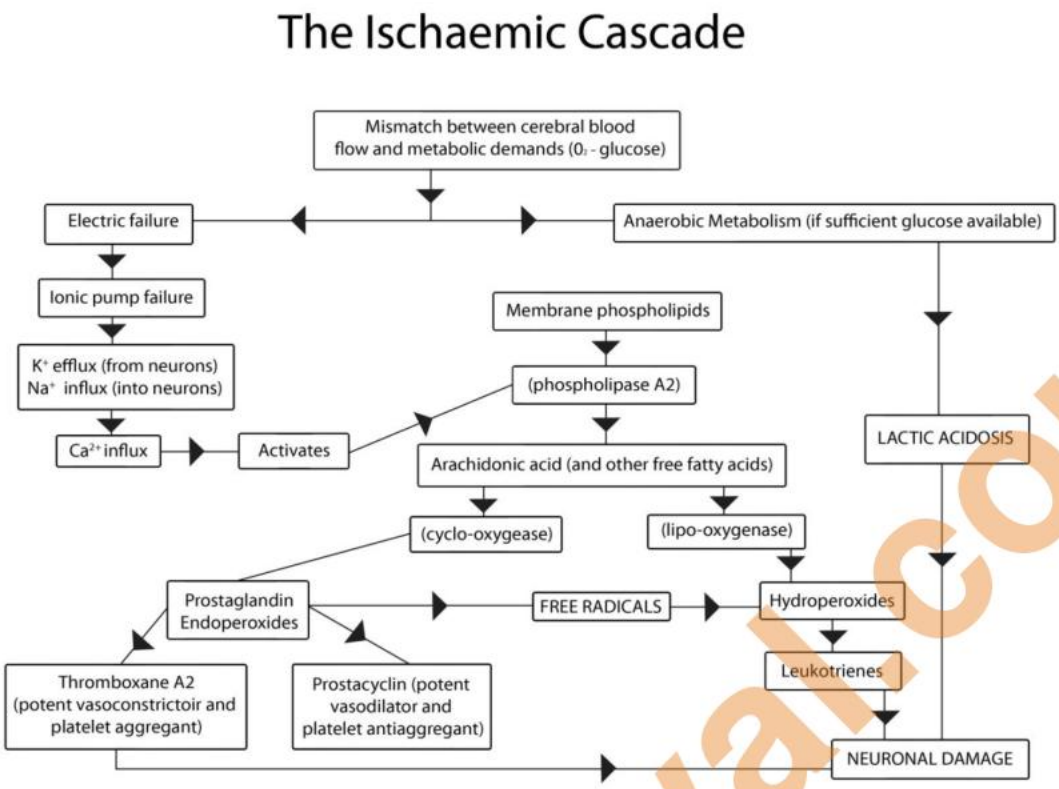
Treatment

1. Controlling high blood pressure (hypertension)
2. Lowering the amount of cholesterol and saturated fat in your diet
3. Quitting tobacco use
4. Controlling diabetes
5. Eating a diet rich in fruits and vegetables
6. Exercising regularly
7. Drinking alcohol

Preventive medications

1. Anti-platelet drugs
2. Anticoagulants
3. Thrombolytics
4. Surgery to remove clot

Pathophysiology



Depression

Depression is classified as a mood disorder. It may be described as feelings of sadness, loss, or anger that interfere with a person's everyday activities.

Types of Depression

1. Major Depression- Severe type of depression
2. Persistent Depressive Disorder- If the depression last for more than 2 year
3. Bipolar Mania- The depressive person sometimes become violent and sometime calm.
4. Seasonal Affective Disorder (SAD)- is a period of major depression that most often happens during the winter months, when the days grow short and you get less and less sunlight. It also occurs in summer.
5. Psychotic Depression- This type of depression occurs when person suffers from psychosis
6. Peripartum (Postpartum) Depression- This depression occurs in female just before the childbirth.
7. Premenstrual Dysphoric Disorder: Before the start of menstrual cycle some female feels depression.
8. Situational' Depression- Some people feels depression in a certain condition.

Cause

- **Family history.** You're at a higher risk for developing depression if you have a family history of depression or another mood disorder.
- **Early childhood trauma.** Some events impact the way that body reacts to fear and stressful situations.
- **Brain structure.** There's a greater risk for depression if the frontal lobe of your brain is less active.
- **Medical conditions.** Certain conditions may put you at higher risk, such as chronic illness, insomnia, chronic pain,.
- **Drug use.** A history of drug or alcohol misuse can impact your risk.
- **Neurotransmitter:** Deficiency of Serotonin and Nor adrenaline is the major cause of depression.
- **Life events.** These include bereavement, divorce, work issues, relationships with friends and family, financial problems

Symptoms

1. **Mood:** anger, aggressiveness, irritability, anxiousness, restlessness
2. **Emotional:** feeling empty, sad, hopeless
3. **Behavioral:** loss of interest, no longer finding pleasure in favorite activities, feeling tired easily, thoughts of suicide, drinking excessively, using drugs, engaging in high-risk activities
4. **Sexual:** reduced sexual desire, lack of sexual performance
5. **Cognitive:** inability to concentrate, difficulty completing tasks, delayed responses during conversations
6. **Sleep:** insomnia, restless sleep, excessive sleepiness, not sleeping through the night
7. **Physical:** fatigue, pains, headache, digestive problems

Diagnosis

There is no specific diagnosis test for test of depression and doctors only rely on sets of question which are asked to the patient

Pathophysiology

Due to various reasons, the level of Serotonin and nor adrenaline level decreases in the brain- and their metabolism also starts in the brain thus leading to reduction of level of both neurotransmitter, this causes the depression.

Treatment

- **Support**: ranging from discussing practical solutions and contributing stresses, to educating family members.
- **Psychotherapy**, also known as talking therapies.
- **Drug treatment**, specifically antidepressants- Imipramine, Tolcapone.
- **Electroconvulsive therapy**, Severe cases of depression that have not responded to drug treatment may benefit from electroconvulsive therapy (ECT)

Schizophrenia

Schizophrenia is a psychosis, a type of mental illness in which a person cannot tell what is real from what is imaginary. It is also known as split mind that person split from the society.

Cause

The exact cause is unclear but it is a mixture of genetic, environmental, neurodevelopmental factors.

1. Genetics: in first-degree relative chance of occurring of this disease is 10%.
2. Environmental factor: maternal viral infection, High blood pressure during pregnancy.
3. Neurodevelopment: During the development of foetus during pregnancy due to various reasons the damage may occur in some part of brain, this may cause schizophrenia.

Theory to explain schizophrenia

Ecological model: This model is related to the surrounding, social, culture, economic and in this model economic model is quiet important, because crisis of money leads to lot of problem in family and society.

Developmental model

Foetal model: Thus if any injury or structural problem arise during development of foetus in pregnancy may lead to schizophrenia.
Childhood development: Thus, sometime during the childbirth and in young age some injury occurs in child which late may develop in schizophrenia.
Genetic model: it has been observed that around 10% chances are there that schizophrenia can be transferred into children form their parents.
Neurotransmitter Theory: this theory states that there may be increased or deregulation of dopamine level in mesolimbic/mesocortical area of brain.
Vulnerability Theory: This theory suggests that attack of schizophrenia may start due to the exposure to some specific situation such as war, riot, illness, flood, fire.

Symptoms

+ve Symptoms- which can be treated by drugs

1. Delusion- false belief
2. Hallucination-false perception, specially of voice
3. Thought disorder
4. Behaviour disorder
5. Speech disorder

-ve Symptoms-which cant be treated by drugs

1. Loss of normal life
2. Loss of expression on face
3. Withdrawl from social contact
4. Lack of personal hygiene
5. Loss of motivation

Diagnosis

1. **Physical exam**
2. **Tests and screenings: CT scan and MRI**
3. **Psychiatric evaluation:** a person has to show at least two of the following symptoms most of the time for a month, and some mental disturbance over 6 months:
    - Delusions (false beliefs that the person won't give up, even when they get proof that they're not true)
    - Hallucinations (hearing or seeing things that aren't there)
    - Disorganized speech and behaviour
    - Catatonic or coma-like daze
    - Bizarre or hyperactive behaviour

Treatment

1. **Medications:** Drugs reducing the level of dopamine in the body- Chlorpromazine, Haloperidol
2. **Psychological Therapy**
    - **Individual therapy**
    - **Social skills training.** This focuses on improving communication and social interactions and improving the ability to participate in daily activities.
    - **Family therapy.** This provides support and education to families dealing with schizophrenia.
    - **Vocational rehabilitation and supported employment.** This focuses on helping people with schizophrenia prepare for, find and keep jobs.
3. **Electroconvulsive therapy**

    For adults with schizophrenia who do not respond to drug therapy, electroconvulsive therapy (ECT) may be considered.

Alzheimer's disease

Alzheimer's disease is a progressive disorder that causes brain cells to waste away (degenerate) and dies. Alzheimer's disease is the most common cause of dementia — a continuous decline in thinking, behavioural and social skills that disrupts a person's ability to function independently.

Alzheimer's disease causes

Experts haven't determined a single cause of Alzheimer's disease but they have identified certain risk factors, including:

1. **Age:** Most people who develop Alzheimer's disease are 65 years of age or older.
2. **Family history:** If you have an immediate family member who has developed the condition, you're more likely to get it.
3. **Genetics:** Certain genes have been linked to Alzheimer's disease.
4. **Protein:**
    - **Plaques-** Beta-amyloid is a leftover fragment of a larger protein. When these fragments cluster together, they appear to have a toxic effect on neurons and to disrupt cell-to-cell communication. These clusters form larger deposits called amyloid plaques, which also include other cellular debris.
    - **Tangles-** Tau proteins play a part in a neuron's internal support and transport system to carry nutrients and other essential materials. In Alzheimer's disease, tau proteins change shape and organize themselves into structures called neurofibrillary tangles. The tangles disrupt the transport system and are toxic to cells.

Symptoms

Early symptom of Alzheimer

1. Difficulty remembering newly learned information.
2. mood and behavior changes

Late symptoms

1. memory loss affecting daily activities, such as an ability to keep appointments
2. trouble with familiar tasks, such as using a microwave
3. difficulties with problem-solving
4. trouble with speech or writing
5. becoming disoriented about times or places
6. decreased judgment
7. decreased personal hygiene
8. mood and personality changes
9. withdrawal from friends, family, and community
10. Multitasking is especially difficult, and it may be challenging to manage finances, balance checkbooks and pay bills on time.
11. The ability to make reasonable decisions and judgments in everyday situations will decline.

Stages of Alzheimer

Stage 1. There are no symptoms at this stage but there might be an early diagnosis based on family history.

Stage 2. The earliest symptoms appear, such as forgetfulness.

Stage 3. Mild physical and mental impairments appear, such as reduced memory and concentration. These may only be noticeable by someone very close to the person.

Stage 4. Alzheimer's is often diagnosed at this stage, but it's still considered mild. Memory loss and the inability to perform everyday tasks is evident.

Stage 5. Moderate to severe symptoms require help from loved ones or caregivers.

Stage 6. At this stage, a person with Alzheimer's may need help with basic tasks, such as eating and putting on clothes.

Stage 7. This is the most severe and final stage of Alzheimer's. There may be a loss of speech and facial expressions.

Pathophysiology

Due to various reasons the degradation of cholinergic neuron takes place in brain. Thus the level of acetylcholine is reduced in brain and meanwhile the acetylcholinsterase enzyme metabolises the acetylcholine thus very less amount of acetylcholine remains in the brain, thus this reduction of acetylcholine level in brain lead to Alzheimer disease.

Diagnosis

A diagnosis of Alzheimer's disease is based on tests your doctor administers to assess memory and thinking skills.

Physical and neurological exam

- Reflexes
- Muscle tone and strength
- Ability to get up from a chair and walk across the room
- Sense of sight and hearing
- Coordination
- Balance

Lab tests

- To detect the level of acetylcholine in the brain.

Brain imaging

- **Magnetic resonance imaging (MRI).** MRI uses radio waves and a strong magnetic field to produce detailed images of the brain.
- **Computerized tomography (CT).** A CT scan, a specialized X-ray technology, produces cross-sectional images (slices) of your brain.
- **Amyloid PET imaging** can measure the burden of amyloid deposits in the brain.
- **Tau Pet imaging,** which measures the burden of neurofibrillary tangles in the brain, is only used in research.

Treatment

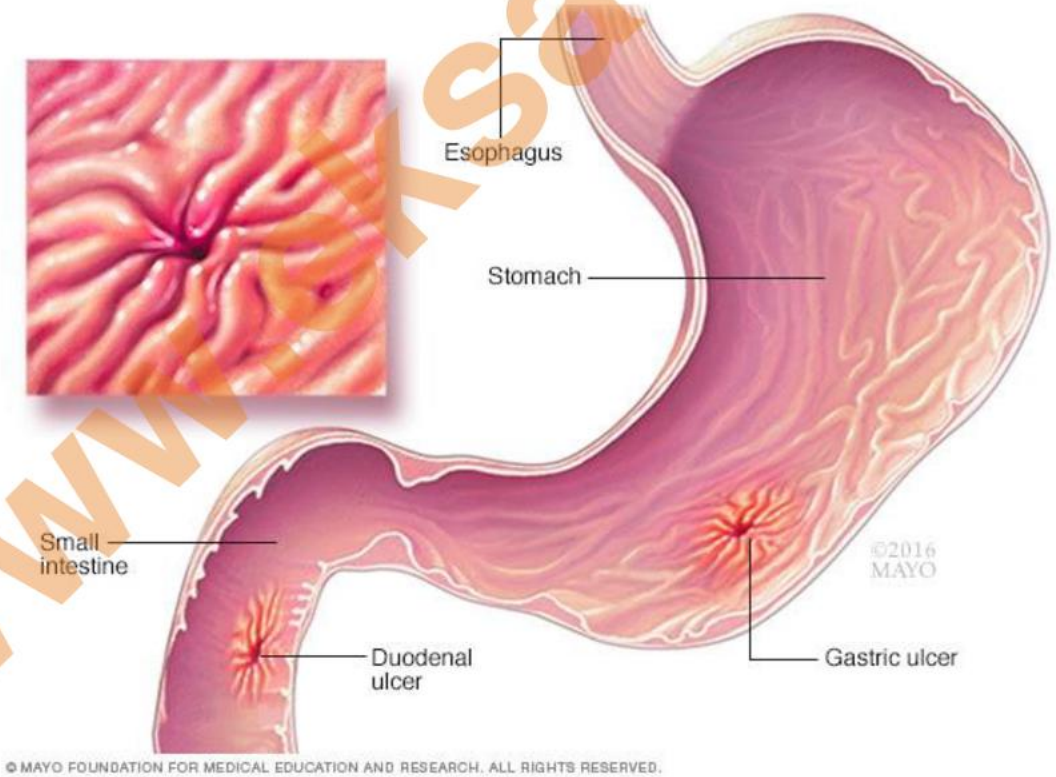
Cholinesterase inhibitors: donepezil, rivastigmine
Alternate treatment: Omega-3 fatty acids**,** Curcumin**,** Ginkgo, Vitamin E

Peptic Ulcers

Peptic ulcers are open injury that develop on the inside lining of your stomach and the upper portion of your small intestine. The most common symptom of a peptic ulcer is stomach pain.

Peptic ulcer is the combination of gastric ulcer and duodenal ulcer.

- **Gastric ulcers** that occur on the inside of the stomach
- **Duodenal ulcers** that occur on the inside of the upper portion of your small intestine (duodenum)



Causes

- *Helicobacter pylori* (*H. pylori*), a type of bacteria that can cause a stomach infection and inflammation
- frequent use of aspirin (Bayer), ibuprofen (Advil), and other anti-inflammatory drugs (risk associated with this behavior increases in women and people over the age of 60)

- smoking
- drinking too much alcohol
- radiation therapy
- stomach cancer
- Smoking cigarettes
- Eat spicy foods

Symptoms

- Burning stomach pain
- Feeling of fullness, bloating or belching
- Fatty food intolerance
- Heartburn
- Nausea
- Vomiting or vomiting blood — which may appear red or blackDark blood in stools, or stools that are black or tarry
- Unexplained weight loss
- Appetite changes

Diagnosis

Two types of tests are available to diagnose a peptic ulcer.

- **Endoscopy:** This test allows doctor to pass a thin, bendy tube down your throat and into your stomach and small intestine. The tube has a camera at the end so she can check the injury of ulcers.
- **Bacterial identification:** Test is performed for the identification of *H.pylori* bacteria

Treatment

- Antibiotic medications to kill H. pylori: Amoxicillin, metronidazole
- Medications that block acid production and promote healing: omeprazole, pantoprazole
- Medications to reduce acid production: Ranitidine, famotidine
- Antacids that neutralize stomach acid: Sodium bicarbonate, Magnesium Hydroxide
- Medications that protect the lining of your stomach and small intestine: Sucralfate, Misopristol.

Iron deficiency

Anaemia is a decrease in the total amount of red blood cells (RBCs) or haemoglobin in the blood

Haemoglobin normal results vary, but in general are:

Male: 13.8 to 17.2 gm/dL

Female: 12.1 to 15.1 gm/dL

Iron deficiency anemia occurs due to shortage of iron in the body. Without enough iron, your body can't produce enough haemoglobin in red blood cells that enables them to carry oxygen. As a result, iron deficiency anemia may leave you tired and short of breath.

Causes

Iron deficiency occurs when the rate of loss or use of iron is more than its rate of absorption and use. The reasons for this are

- Ingesting food which has low iron content.
- Chronic blood loss: Most commonly due to excessive menstruation or bleeding into or from the gut as a result of a peptic ulcer, gastritis, haemorrhoids or in children, worm infestation.
- Increased use of iron: In pregnancy, due to the growth of the foetus or children undergoing rapid growth.
- Decreased absorption of iron due to:
    - partial or total removal from the stomach;
    - lack of stomach acid;
    - chronic diarrhoea; or
    - malabsorption.

Signs and symptoms

Initially, iron deficiency anemia can be so mild that it goes unnoticed. But as the body becomes more deficient in iron and anemia worsens, the signs and symptoms are as follow:

- Extreme fatigue
- Weakness
- Pale skin
- Chest pain, fast heartbeat or shortness of breath
- Headache, dizziness or lightheadedness
- Cold hands and feet
- Inflammation or soreness of your tongue
- Brittle nails
- Unusual cravings for non-nutritive substances, such as ice, dirt or starch
- Poor appetite, especially in infants and children with iron deficiency anemia

Diagnosis

To diagnose iron deficiency anemia, these tests are done to look for:

- **Red blood cell size and color.** With iron deficiency anemia, red blood cells are smaller and lighter in colour than normal.
- **Hematocrit.** This is the percentage of your blood volume made up by red blood cells. Normal levels are generally between 34.9 and 44.5 percent for adult women and 38.8 to 50 percent for adult men.
- **Hemoglobin.** Lower than normal hemoglobin levels indicate anemia. The normal hemoglobin range is generally defined as 13.5 to 17.5 grams (g) of hemoglobin per deciliter (dL) of blood for men and 12.0 to 15.5 g/dL for women.
- **Ferritin.** This protein helps store iron in your body, and a low level of ferritin usually indicates a low level of stored iron

Treatment

1. Take iron rich diet
    - Meat, especially beef and liver
    - Poultry — chicken livers are packed with iron
    - Fish and shellfish, especially oysters
    - Leafy greens, like kale, spinach, and broccoli
    - Beans and peas
    - Iron-enriched breads, pastas, and cereals
2. Take iron tablets
3. Take iron tablets with vitamin C: Vitamin C improves the absorption of iron.
4. Blood transfusions
5. Intravenous iron therapy

Megaloblastic anemia

Megaloblastic anaemia is a condition in which the bone marrow produces unusually large, structurally abnormal, immature red blood cells. Generally bigger size RBC is produced in this type of anemia.

Cause

The two most common causes of megaloblastic anemia are deficiencies of vitamin B-12 or folic acid. These two nutrients are necessary for producing normal size red blood cells.

Vitamin B-12 Deficiency
Vitamin B-12 is a nutrient found in some foods like meat, fish, eggs, and milk. Some people can't absorb enough vitamin B-12 from their food, leading to megaloblastic anemia. Megaloblastic anemia caused by vitamin B-12 deficiency is referred to as pernicious anemia. Vitamin B-12 deficiency is most often caused by the lack of a protein in the stomach called "intrinsic factor." Without intrinsic factor, vitamin B-12 can't be absorbed, regardless of how much you eat.

Folic Acid Deficiency
Folic acid is another nutrient that's important for the development of normal size red blood cells. Folate is found in foods like beef liver, spinach, and Brussels sprouts. Folate deficiency can also be caused by chronic alcohol abuse, since alcohol interferes with the body's ability to absorb folic acid. Pregnant women are more likely to have folate deficiency, because of the high amounts of folate needed by the developing fetus.

Pathophysiology

Due to deficiency of folic acid and vitamin B_{12}, the DNA synthesis in the red blood cell production is inhibited. When DNA synthesis is inhibited, the cell cycle cannot progress from the G2 growth stage to the mitosis (M) stage. This leads to continuing RBC growth without division, resulting in formation of RBC of bigger size.

Symptom of megaloblastic anemia

- Fatigue
- shortness of breath
- muscle weakness
- abnormal paleness of the skin
- glossitis (swollen tongue)
- loss of appetite/weight loss
- diarrhea
- nausea
- fast heartbeat
- smooth or tender tongue
- tingling in hands and feet
- numbness in extremities

Diagnosis

- Complete blood count (CBC): This test measures the different parts of your blood. It checks the number and appearance of your red blood cells. They will appear larger and underdeveloped if you have megaloblastic anemia.
- Schilling test: The Schilling test is a blood test that evaluates your ability to absorb vitamin B-12.

- Vitamin B-12 Level test: level of Vitamin B_{12} is identified.

Treatment

Vitamin B-12 Deficiency

1. Monthly injections of vitamin B-12
2. Oral supplements of vitamin B-12
3. Diet
    - eggs
    - chicken
    - fortified cereals (especially bran)
    - red meats (especially beef)
    - milk
    - shellfish

Folic acid Deficiency

1. Oral supplements of vitamin B-12 and folic acid.
2. Diet
    - oranges
    - leafy green vegetables
    - peanuts
    - lentils
    - enriched grains

Sickle cell anemia

Sickle cell anemia is a genetically transferred form of anemia, in which the red blood cells become rigid and sticky and are shaped like sickles. These irregularly shaped cells can get stuck in small blood vessels, which can slow or block blood flow and oxygen supply to parts of the body.

Causes

Sickle cell anemia is caused by a mutation in the gene that tells your body to make the red, iron-rich compound that gives blood its red color (hemoglobin). Hemoglobin allows red blood cells to carry oxygen from your lungs to all parts of your body. In sickle cell anemia, the abnormal hemoglobin causes red blood cells to become rigid, sticky and misshapen.
The sickle cell gene is passed from generation to generation in a pattern of inheritance called autosomal recessive inheritance. This means that both the mother and the father must pass on the defective form of the gene for a child to be affected.

Symptoms

1. **Anemia.** Sickle cells break apart easily and die, leaving you without enough red blood cells. Red blood cells usually live for about 120 days before they need to be replaced. But sickle cells usually die in 10 to 20 days, leaving a shortage of red blood cells (anemia).
2. **Episodes of pain:** Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints. Pain can also occur in your bones.
3. **Painful swelling of hands and feet.** The swelling is caused by sickle-shaped red blood cells blocking blood flow to the hands and feet.
4. **Frequent infections.** Sickle cells can damage an organ that fights infection (spleen), leaving you more vulnerable to infections.
5. **Delayed growth.** Red blood cells provide your body with the oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.

6. **Vision problems.** Tiny blood vessels that supply your eyes may become plugged with sickle cells. This can damage the retina — the portion of the eye that processes visual images, leading to vision problems.

Diagnosis

A blood test can check for hemoglobin S — the defective form of hemoglobin that underlies sickle cell anemia.

Tests to detect sickle cell genes before birth

Sickle cell disease can be diagnosed in an unborn baby by sampling some of the fluid surrounding the baby in the mother's womb (amniotic fluid) to look for the sickle cell gene.

Treatment

No. As of today, there's no cure for sickle cell anemia.

1. **Bone marrow transplant:** A bone marrow transplant, also called a stem cell transplant, involves replacing bone marrow affected by sickle cell anemia with healthy bone marrow from a donor.
2. **Blood transfusions:** In a red blood cell transfusion, red blood cells are removed from a supply of donated blood, and then given intravenously to a person with sickle cell anemia.
3. **Medications**
4. **Antibiotics**
5. **Pain-relieving medications.**
6. **Vaccinations to prevent infections:** Childhood vaccinations are important for preventing disease in all children. They're even more important for children with sickle cell anemia because their infections can be severe.
7. **Gene therapy**

Thalassemia

Thalassemia is a blood disorder passed down through families (inherited) in which the body characterized by less hemoglobin and fewer red blood cells in your body than normal.

Causes

Hemoglobin is made of two proteins:

- Alpha globin
- Beta globin

Thalassemia occurs when there is a defect in a gene that helps control production of one of these proteins.

There are two main types of thalassemia:

- Alpha thalassemia occurs when a gene or genes related to the alpha globin protein are missing or changed (mutated).
- Beta thalassemia occurs when similar gene defects affect production of the beta globin protein.

Symptoms

- Fatigue
- Weakness
- Pale or yellowish skin
- Facial bone deformities
- Slow growth
- Abdominal swelling
- Dark urine

Diagnosis

If you are suffering from thalassemia, blood tests may reveal:

- A low level of red blood cells
- Smaller than expected red blood cells
- Pale red blood cells
- Red blood cells that are varied in size and shape
- Red blood cells with uneven hemoglobin distribution, which gives the cells a bull's-eye appearance under the microscope

Treatment

Treatment for thalassemia depends on which type you have and how severe it is.

1. Frequent blood transfusions
2. Eat a healthy diet
3. Avoid infections
4. bone marrow transplant
5. medications and supplements

Haemophilia

Haemophilia is a rare disorder in which your blood doesn't clot normally because it lacks sufficient blood-clotting proteins (clotting factors).

Causes

It's caused by a defect in the gene that determines how the body makes factors VIII, IX, or XI. These genes are located on the X chromosome, so the transfer of this defected chromosome into child may lead to haemophilia.

Types

The three forms of haemophilia are haemophilia A, B, and C.

- Haemophilia A is the most common type of haemophilia, and it's caused by a deficiency in factor VIII.
- Haemophilia B, which is also called Christmas disease, is caused by a deficiency of factor IX.
- Haemophilia C is a mild form of the disease that's caused by a deficiency of factor XI.

Symptoms

Mild haemophilia: Bleeding will continue when the person met a trauma
Severe haemophilia: People with a severe deficiency may bleed for no reason. This is called "spontaneous bleeding."
Spontaneous bleeding can cause the following:

- blood in the urine
- blood in the stool
- deep bruises
- large, unexplained bruises
- excessive bleeding
- bleeding gums
- frequent nosebleeds
- pain in the joints
- tight joints
- irritability (in children)

Diagnosis

A clotting-factor test in the blood is done in order to find out amount of clotting factor present in the blood. The sample is then graded to determine the severity of the factor deficiency:

- Mild haemophilia is indicated by a clotting factor in the plasma that's between 5 and 40 percent.
- Moderate haemophilia is indicated by a clotting factor in the plasma that's between 1 and 5 percent.
- Severe haemophilia is indicated by a clotting factor in the plasma of less than 1 percent.

Treatment

1. **Desmopressin (DDAVP):** In mild haemophilia, this hormone can stimulate your body to release more clotting factor.
2. Infusing blood with donor clotting factors.
3. **Clot-preserving medications (anti-fibrinolytics):** These medications help prevent clots from breaking down.
4. **Fibrin sealants:** These medications can be applied directly to wound sites to promote clotting and healing.
5. **First aid for minor cuts:** Using pressure and a bandage will generally take care of the bleeding.

Diabetes

Diabetes mellitus, commonly known as diabetes, is a metabolic disease in which the sugar level in the blood rises above the normal level. This disease is related to hormone insulin.

Normal sugar level in blood:

Category	Fasting (mg/dl)	Post Prandial (PP) (After 2 Hour of eating) (mg/dl)
Normal	70-100	100-140
Pre-Diabetic	100-125	140-180
Diabetic	More than 125	180 above

Types

1. **Type 1 diabetes** is also called insulin-dependent diabetes.
2. **Type 2 diabetes** is also called Non-insulin-dependent diabetes.
3. **Gestational diabetes** is high blood sugar during pregnancy.

Symptoms of diabetes

- Increased hunger
- Increased thirst
- Weight loss
- Frequent urination
- Nerve damage (Diabetic neuropathy)
- Kidney damage (Diabetic nephropathy)
- Eye damage (Diabetic retinopathy)
- Extreme fatigue
- Sores that don't heal

Cause

Type 1 diabetes: It is an autoimmune condition. It's caused by the body attacking its own pancreas with antibodies. In people with type 1 diabetes, the damaged pancreas doesn't make insulin.

Type 2 diabetes: Type 2 diabetes stems from a combination of genetics and lifestyle factors. Being overweight or obese increases your risk too. Carrying extra weight, especially in your belly, makes your cells more resistant to the effects of insulin on your blood sugar or pancreas is unable to produce insulin according to body requirement.

Gestational diabetes: Gestational diabetes is the result of hormonal changes during pregnancy. The placenta produces hormones that make a pregnant woman's cells less sensitive to the effects of insulin. This can cause high blood sugar during pregnancy.

Risk

Your risk for type 2 diabetes increases if you:

- are overweight
- are age 45 or older
- have a parent or sibling with the condition
- aren't physically active
- have had gestational diabetes
- have prediabetes
- have high blood pressure, high cholesterol, or high triglycerides

Diagnosis

Sugar level estimation test is done in the body. If the level is above normal then that person can be categorised as prediabetic or diabetic.

Treatment

Type 1 diabetes

Insulin is the main treatment for type 1 diabetes, because our body is unable to produce insulin by its own so it is given from outside in the form of injection.
There are four types of insulin that are most commonly used. They're differentiated by how quickly they start to work, and how long their effects last:

- Rapid-acting insulin starts to work within 15 minutes and its effects last for 3 to 4 hours.
- Short-acting insulin starts to work within 30 minutes and lasts 6 to 8 hours.
- Intermediate-acting insulin starts to work within 1 to 2 hours and lasts 12 to 18 hours.
- Long-acting insulin starts to work a few hours after injection and lasts 24 hours or longer.

Type 2 diabetes

1. Life style modification
    - Eating a diet high in fresh, nutritious foods, including whole grains, fruits, vegetables, lean proteins, low-fat dairy, and healthy fat sources, such as nuts.
    - Avoiding high-sugar foods that provide empty calories.
    - Avoid from drinking excessive amounts of alcohol.
    - Engaging in at least 30 minutes exercise a day on at least 5 days of the week.
    - Recognizing signs of low blood sugar when exercising, including dizziness, confusion, weakness, and profuse sweating.
2. Medicines- Metformin, gilpizide

Thyroid diseases
Goitre

The term "goitre" simply refers to the abnormal enlargement of the thyroid gland. The thyroid gland is a small butterfly-shaped gland in the neck, just in front of the windpipe (trachea). It produces thyroid hormones, which helps to regulate the body's metabolism (the process that turns food into energy). A swelling of thyroid gland can further lead to a swelling of the neck or larynx (voice box). Goitre is also known as Iodine deficiency disorder as Iodine deficiency.

Types of goitre: There are two different types of goitre:

- **Diffuse small goitre:** where entire thyroid gland enlarges to a larger size and feels smooth to the touch
- **Nodular goitre:** where certain sections, or "nodules", of thyroid gland enlarge and feel lumpy to the touch.

Symptoms

- Swelling of the thyroid gland,
- Swelling of neck which causes a lump to develop in the throat.
- Breathing difficulties
- Cough
- Hoarseness of voice
- Difficulty in swallowing specially with solid foods.

Cause

Deficiency of Iodine

Pathophysiology

Due to deficiency of iodine, the thyroid gland does not produce enough thyroid hormones, the deficiency of thyroid hormones occurs in the body, the body will stimulate it to produce more. This can cause the thyroid gland to enlarge and form a lump in the neck.

Diagnosis

Physical examination: Examination of the swelling will help the physician to diagnose goitre.

- A diffuse small goitre, where entire thyroid gland swells up
- A nodular goitre, where certain sections, or "nodules" of thyroid gland swell up.
- Levels of thyroid hormone.
- Ultrasound

Treatment

- Iodine supplement
- Surgery
- Hormone therapy: The synthetic hormone (levothyroxine) is given orally

Hyperthyroidism

Hyperthyroidism (overactive thyroid) occurs when your thyroid gland produces too much of the hormone thyroxine. Hyperthyroidism can accelerate your body's metabolism, causing unintentional weight loss and a rapid or irregular heartbeat.

Causes

- **Graves' disease:** Graves' disease is an autoimmune disorder in which antibodies produced by your immune system stimulate your thyroid to produce too much T4. It's the most common cause of hyperthyroidism.
- **Plummer's disease:** This form of hyperthyroidism occurs when one or more part of your thyroid produce too much T4.
- **Thyroiditis:** The inflammation can cause excess thyroid hormone stored in the gland to leak into your bloodstream.

Symptoms

- Unintentional weight loss, even when your appetite and food intake stay the same or increase
- Rapid heartbeat (tachycardia) — commonly more than 100 beats a minute
- Irregular heartbeat (arrhythmia)
- Pounding of your heart (palpitations)
- Increased appetite
- Nervousness, anxiety and irritability
- Tremor — usually a fine trembling in your hands and fingers
- Sweating
- Changes in menstrual patterns
- Increased sensitivity to heat
- Changes in bowel patterns, especially more frequent bowel movements
- Fatigue, muscle weakness
- Difficulty sleeping
- Skin thinning
- Fine, brittle hair
- Red or swollen eyes

Diagnosis

- **Medical history and physical exam:** During the exam your doctor may try to detect a slight tremor in your fingers when they're extended, overactive reflexes, eye changes and warm, moist skin.
- **Blood tests:** Blood tests that measure thyroxine and thyroid-stimulating hormone (TSH) can confirm the diagnosis. High level tells about hyperthyroidism.

Treatment

Radioactive iodine: Taken by mouth, radioactive iodine is absorbed by your thyroid gland, where it causes the gland to shrink.

Anti-thyroid medications: These medicines prevent the formation and release of T3 and T4 hormones.

Surgery: To remove some part of thyroid gland, when the tissues are removed lee formation and release of thyroid hormones will take place.

Disorders of sex hormones

The testes in men produces testosterone and the ovaries in women produce progesterone and estrogen. Each of these glands is controlled by the pituitary gland which in turn is controlled by the hypothalamus. Imbalance in production and release of these hormones will lead to the sexual disease.

Females

- Polycystic ovary syndrome
- Hirsutism

Male

- Hypogonadism
- Gynecomastia

Polycystic ovary syndrome

Polycystic ovary syndrome (PCOS) is a condition that affects a woman's hormone levels. Women with PCOS produce higher-than-normal amounts of male hormones. This hormone imbalance causes them to skip menstrual periods and makes it harder for them to get pregnant. PCOS also causes hair growth on the face and body, and baldness.

Cause

- The high levels of male hormones prevent the ovaries from producing female hormones and making eggs normally.
- It is transferred from one generation to another.

Symptoms

- **Irregular periods** (Dysmenorrhea)**:** A lack of ovulation prevents the uterine lining from shedding every month.
- **Amenorrhea:** Absence of menstrual cycle.
- **Heavy bleeding:** The uterine lining builds up for a longer period of time, so the periods you do get can be heavier than normal.
- **Hair growth:** More than 70 percent of women with this condition grow hair on their face and body — including on their back, belly, and chest. Excess hair growth is called hirsutism.
- **Acne:** Male hormones can make the skin oilier than usual and cause breakouts on areas like the face, chest, and upper back.
- **Weight gain:** Up to 80 percent of women with PCOS are overweight or obese.
- **Male-pattern baldness:** Hair on the scalp gets thinner and fall out.
- **Darkening of the skin:** Dark patches of skin can form in body creases like those on the neck, in the groin, and under the breasts.
- **Headaches:** Hormone changes can trigger headaches in some women.

Diagnosis

- High androgen levels
- Irregular menstrual cycles
- Cysts in the ovaries

Treatment

- Estrogen and progestin
- Clomiphene
- Surgery

Hirsutism

Hirsutism (HUR-soot-iz-um) is a condition of unwanted, male-pattern hair growth in women. Hirsutism results in excessive amounts of dark, course hair on body areas where men typically grow hair — face, chest and back.

Symptoms

- Excessive growth of hair on the body of women.
- Deepening voice
- Balding
- Acne

Causes

- Polycystic ovary syndrome
- Tumors
- Medications.
- Family history

Pathophysiology

At puberty, a girl's ovaries begin to produce a mixture of female and male sex hormones, causing hair to grow in the armpits and pubic area. Hirsutism can occur if the ratio of male and female hormone production is disturbed and female ovary start secreting more male hormones, then this leads to the development of hair on the female body.

Diagnosis

- Testosterone level
- An ultrasound or a CT scan to check your ovaries and adrenal glands for tumors or cysts.

Treatments

- Estrogen and progestin
- Anti-androgens medicine (spironolactone)
- Laser therapy

Hypogonadism

Hypogonadism is a disease when your sex glands (testes) produce little or no sex hormones (testosterone).

Types

There are two types of hypogonadism

- Primary
- Central

Primary hypogonadism

Primary hypogonadism means that your testes are unable to produce normal amount of testosterone.

Central (secondary) hypogonadism

In central hypogonadism, the problem lies in your brain. Your hypothalamus and pituitary gland, which control your testes, aren't working properly.

Cause

Primary hypogonadism

- autoimmune disorders, such as Addison's disease
- genetic disorders, such as Turner syndrome and Klinefelter syndrome
- severe infections, especially mumps involving your testicles
- liver and kidney diseases
- undescended testes
- hemochromatosis, which happens when your body absorbs too much iron
- radiation exposure
- surgery on your sexual organs

Central hypogonadism

- genetic disorders, such as Kallmann syndrome (abnormal hypothalamic development)
- infections, including HIV
- pituitary disorders
- inflammatory diseases, including sarcoidosis, tuberculosis, and histiocytosis
- obesity
- rapid weight loss
- nutritional deficiencies
- use of steroids or opioids
- brain surgery
- radiation exposure
- injury to your pituitary gland or hypothalamus
- A tumor in or near your pituitary gland

Symptoms of hypogonadism

Symptoms that may appear in males include:

- loss of body hair
- muscle loss
- abnormal breast growth
- reduced growth of penis and testicles
- erectile dysfunction
- osteoporosis
- low or absent sex drive
- infertility
- fatigue
- hot flashes
- difficulty concentrating
- less sperm count

Diagnosis

- **Hormone tests-** Check the level of follicle-stimulating hormone (FSH) and luteinizing hormone.

- **Imaging test**- An ultrasound, CT-scan, MRI scan.

Treatment

Testosterone is a male sex hormone. Testosterone replacement therapy is a widely used treatment for hypogonadism in males. You can get testosterone replacement therapy by:

- injection
- patch
- gel
- lozenge

Injections of a gonadotropin-releasing hormone may trigger puberty or increase your sperm production.

Hypergonadism

Hypergonadism is a condition in which testes overproduce hormones (testosterone).

Cause

- central nervous system abnormalities
- rare genetic disorders
- tumors in the pituitary gland or brain
- tumors in an ovary or testis
- adrenal gland disorder
- severe hypothyroidism (underactive thyroid)
- severe infections
- surgery
- injury to testes

Symptoms

- early growth spurts
- mood swings
- acne
- a lower voice
- more muscle mass
- increased sex drive
- spontaneous erections and nocturnal emissions

Diagnosis

- **Hormone tests-** Check the level of follicle-stimulating hormone (FSH) and luteinizing hormone.
- **Imaging test**- An ultrasound, CT-scan, MRI scan.

Treatment

Treatment for hypergonadism can be an intensive process as it is difficult to lower the hormone levels as compared to increasing them and patients are treated by anti-testosterone drugs and surgery.