

Pathophysiology (Unit-3)

B. Pharma 2nd semester

UNIT - III

★ Haematological Disease :-

The branch of medical science that deals with the form and structure of blood components and blood-forming tissue is determined termed as Haematology.

- Blood has two major components i.e. plasma and formed elements. Formed elements include the three types of cells present in blood.

1. Red blood cells (RBCs) / Erythrocytes.
2. White blood cells (WBC) / Leukocytes
3. Platelets / Thrombocytes.

- The formation and development of blood cells normally take place in the red bone marrow and under the control of hormones and feedback mechanism that maintains the required amount of blood.

- Common haematological disorders are:

1. Anemia
2. Leukemia
3. Bleeding disorder
4. Blood cancers etc.

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① Iron deficiency :-

Iron deficiency is a Condition characterized by a lack of sufficient iron in the body, which can lead to various health issues, most notably anemia.

Causes:-

1. Inadequate Iron intake: Insufficient dietary intake of iron-rich foods.
2. Increased iron Demand: Higher iron requirements during periods of rapid growth (infancy, adolescence), pregnancy and lactation.
3. Blood Loss: Menstruation, gastrointestinal bleeding, surgeries or trauma.
4. Poor Absorption: Condition like Celiac disease, Crohn's disease or certain medications that interfere with iron absorption.

Symptoms:-

1. Fatigue and Weakness: Due to reduced oxygen transport in blood.
2. Pale Skin: From decreased hemoglobin levels.
3. Shortness of breath: Especially during physical activity.
4. Cold Hands and Feet: Poor circulation.

Diagnosis :-

1. Complete blood Count (CBC): To check hemoglobin and hematocrit levels.
2. Serum ferritin: To measure stored iron in the body.
3. Serum iron and Total Iron Binding Capacity (TIBC): To assess iron transport and binding capacity.

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Treatment :-

1. **Dietary changes:** Increase intake of iron-rich foods like red meat, poultry fish, legumes, and Leafy green vegetables.
2. **Iron Supplements:** Oral Iron Supplements (ferrous Sulfate, ferrous gluconate) or Intravenous iron for Severe Cases.
3. **Treat Underlying Causes:** Address any underlying Conditions Causing blood loss or poor iron absorption.
4. **Vitamin C Intake:** Enhances Iron absorption when taken with iron rich foods or Supplements.

② Megaloblastic Anemia :-

Megaloblastic anemia is a type of anemia characterized by the presence of abnormally large red blood cells (megaloblasts) due to impaired DNA synthesis. often Caused by deficiencies in Vit-B12 or folate.

Causes :-

1. Vit B12 Deficiency :-

- Inadequate dietary intake
- pernicious anemia (autoimmune Condition affecting Vit-B12 absorption).
- Malabsorption disorders (e.g. Celiac disease, Crohn's disease)
- Gastric Surgeries affecting the Stomach or small Intestine.

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2. Folate Deficiency :-

- Poor dietary intake of folate
- Malabsorption disorder
- Increased demand (pregnancy, lactation)
- Certain medications (e.g. Methotrexate, anti convulsants)

Symptoms :-

1. General Anemia Symptoms :-

- Fatigue and weakness
- Dizziness or lightheadedness
- Palor
- Rapid heartbeat
- Shortness of breath

2. Neurological Symptoms (vit-B12 deficiency) :-

- Numbness or tingling in the hand and feet
- Difficulty walking or balance problems
- Cognitive disturbances (Memory loss, Confusion)

3. Gastrointestinal Symptoms :-

- Loss of appetite
- Weight loss
- Sore tongue or mouth

Diagnosis :-

1. Complete blood Count (CBC) : To check for anemia and the presence of large red blood cells.

2. Peripheral blood Smear : To observe the size and Shape of RBC.

3. Serum Vit-B12 and Folate level : To detect deficiencies.

4. Bone Marrow Examination : In Severe Case to observe Megaloblasts in the bone marrow.

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Treatment :-

1. Vit-B12 Supplementation:-

- oral or intramuscular injections of vit-B12.
- Dietary change to include more vitamin B12-rich foods (meat, dairy, eggs).

2. Folate Supplementation:-

- oral folic acid supplements.
- Dietary changes to include more folate-rich foods (leafy green vegetables, legumes, fortified cereals).

3. Treat Underlying Condition:-

Address any malabsorption issues or other contributing factors.

③ Sickle Cell Anemia:-

A inherited blood disorder in which abnormal haemoglobin causes the RBCs to distort and attain sickle-like shape. It is termed as Sickle Cell anemia.

Genetic Cause :-

- Sickle Cell anemia is caused by a mutation in the hemoglobin gene. The mutation leads to the production of hemoglobin S instead of normal hemoglobin (A).
- The mutation results in the formation of sickle-shaped red blood cells under certain conditions (low oxygen levels, dehydration etc).

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- Normal RBC are flexible and round, allowing them to flow easily through blood vessels.
- In sickle cell anemia, the presence of hemoglobin S causes red blood cells to become rigid and shaped like crescent moons or sickles.
- These abnormal cells can clump together and block blood flow in small blood vessels. This blockage leads to pain, tissue damage and various complications.

Sign & Symptoms:-

- | | |
|-----------------------|------------------|
| - Fatigue | - Breathlessness |
| - Rapid heartbeat | - Headache |
| - Cold hands and feet | - Paleness |
| - Chest pain | |

Diagnosis:-

- Diagnosis is typically confirmed by hemoglobin electrophoresis which identifies the presence of hemoglobin C.
- Newborn screening programs in many countries help identify infants with sickle cell disease early, allowing for early intervention and management.

Treatment & Management:-

Pain Management: Pain during sickle cell crises is managed with pain medications, hydration and sometimes hospitalization.

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- **Hydroxyurea**: This medication can help reduce the frequency of Sickle Cell Crises by increasing the production of fetal hemoglobin, which interfere with sickling process.
- **Blood Transfusions**: used to treat Severe anemia and prevent complications such as stroke.
- **Bone Marrow or Stem Cell Transplant**:
Curation option for some patients, but it carries significant risk and is not suitable for everyone.

④ Thalassemia:

Thalassemia is a genetic blood disorder characterized by abnormal hemoglobin production, leading to anemia.

Types: There are two main type of thalassemia based on which part of the hemoglobin molecule is affected.

1. **Alpha thalassemia**: Involve Mutation in the genes that Code for alpha globin chains.
2. **Beta thalassemia**: Involve Mutation in the genes that Code for Beta globin chains.

Symptoms:

It is often asymptomatic or may cause Mild anemia

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- **Moderate to severe:** Can lead to Significant anemia, fatigue, weakness, shortness of breath, jaundice, and in Severe Cases, bone deformities and growth issues in children.

Diagnosis:-

Typically Confirmed through blood test that Measure hemoglobin Levels and examine the Size and Shape of Red blood Cells. Genetic testing Can Identify Specific Mutation.

Treatment:- Treatment depends on the type and Severity.

- **Blood transfusion:** Regular transfusion Can help Maintain hemoglobin Levels in Severe Cases.
- **Iron Chelation therapy:** Necessary to remove excess Iron that accumulates from frequent transfusion.
- **Bone Marrow transplant:** Curative for some patients, especially in Severe Cases of thalassemia Major.

⑤ Hemophilia:-

Hemophilia is a genetic disorder characterized by the inability of blood to clot normally due to a deficiency or absence of specific Clotting proteins.

Types:- There are Several type of hemophilia, the Most Common being.

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- Hemophilia A: Deficiency of clotting factor VIII
- Hemophilia B: Deficiency of clotting factor IX
- Hemophilia C: Deficiency of clotting factor XI (less common)

Symptoms :-

- Excessive bleeding following injuries, surgeries or dental procedure
- Spontaneous bleeding into joints, muscles or other tissue.
- Easy bruising.
- Prolonged bleeding after minor cuts or abrasions.

Diagnosis :-

Diagnosis involves blood test to measure clotting factor level and genetic testing to identify the specific mutation causing hemophilia.

Treatment :-

- Clotting Factor Replacement Therapy: The main treatment involves infusion of clotting factors (factor VIII or IX) to ~~reset~~ restore normal clotting function.
- Desmopressin: In some cases of mild hemophilia A, this medication can stimulate the release of clotting factor VIII.
- Gene Therapy: Emerging as a potential treatment to correct the genetic defects and produce clotting factors within the body.

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★ Endocrine System:-

The endocrine system is a network of gland and organs that produce and release hormones to regulate various functions of the body.

- The endocrine system uses chemical messengers called hormones.
- These hormones are released directly into the bloodstream and carried to target cells or organs throughout the body.
- Key components of the endocrine system include:

1. Glands: e.g. pituitary gland, thyroid gland, adrenal, pancreas, and reproductive gland (ovaries and testes).

2. Hormones: These are molecules that act as chemical messengers. Hormones regulate such as process such as metabolism, growth and development, mood and reproductive cycles.

3. Feedback Mechanisms: Hormone release is often controlled by feedback mechanisms to maintain balance in the body.

4. Functions: The endocrine system plays a crucial role in maintaining homeostasis.

- Metabolism and energy balance.
- Growth and development.
- Reproduction and sexual functions.
- Regulation of blood sugar levels and fluid balance.

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*① Diabetes :-

Diabetes Mellitus is a Chronic Metabolic disorder characterized by high blood sugar levels (hyperglycemia) due to ~~their~~ either insufficient insulin production, insulin resistance, or both.

Types of Diabetes :-

1. Type 1 Diabetes :-

- Autoimmune Condition where the immune system attacks insulin-producing beta cells in the pancreas.
- Requires lifelong insulin therapy.
- Typically diagnosed in children and young adults.

2. Type 2 Diabetes :-

- Result from insulin resistance, where cells fail to respond to insulin properly.
- Can be managed with lifestyle changes, oral medications, and sometimes insulin.
- Linked to obesity and sedentary lifestyle.

3. Gestational Diabetes :-

- Develops during pregnancy and usually resolves after childbirth.
- Increase risk for both the mother and child to develop type 2 diabetes later.

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Symptoms :-

- Excessive thirst (polydipsia) and hunger (polyphagia)
- Frequent urination (polyuria)
- Fatigue and Irritability
- Blurred vision
- Slow wound healing

Management :-

- Type 1 Diabetes: Insulin therapy through injections or pump
- Type 2 Diabetes: Diet and exercise, oral medications (e.g. Metformin) insulin if needed.
- Blood Sugar monitoring
- Healthy diet and regular physical activity
- Avoidance of smoking and excessive alcohol consumption

prevention :-

- maintain a healthy weight
- Engage in regular physical activity
- Eat a balanced diet rich in fruits, vegetables and whole grains.
- Avoid excessive sugar intake and unhealthy fat.

Monitoring :-

- Regular check-ups with healthcare provider
- Monitoring blood sugar levels (Self-Monitoring)

Screening for complications (eye exams, kidney function tests)

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② Thyroid diseases:

Thyroid disease refers to any dysfunctions of the thyroid gland, a butterfly-shaped gland located in the neck responsible for producing hormones that regulate Metabolism.

Types of thyroid disorders:

1. Hypothyroidism:

- Underactive thyroid gland
- Insufficient production of thyroid hormones (T_3 and T_4)
- Symptoms include: fatigue, weight gain, Cold intolerance, dry skin and Constipations.
- Common Causes: autoimmune thyroiditis (Hashimoto's thyroiditis) thyroid surgery or radiation therapy.

2. Hyperthyroidism:

- overactive thyroid gland.
- Excessive production of thyroid hormones
- Symptoms include weight loss, increase appetite, heat intolerance, sweating, palpitations, and nervousness.
- Common Causes: Graves' disease (autoimmune disorder), thyroid nodules or thyroiditis.

3. Thyroid Nodules:

- Abnormal growths or lumps within the thyroid gland.
- May be benign (non-cancerous) or malignant (cancerous)
- often asymptomatic but can cause difficulty swallowing, or visible neck swelling.

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④ Thyroiditis:-

- Inflammation of the thyroid gland.
- Can be Caused by autoimmune Conditions, viral infections, or postpartum changes.
- Types include Hashimoto's thyroiditis (chronic autoimmune thyroiditis) and Subacute thyroiditis.

Diagnosis :-

- Blood tests to measure thyroid hormone level (TSH , T_3 , T_4).
- Imaging Studies (Ultrasound, thyroid scan), to assess gland size and nodules.
- Fine-needle aspiration (FNA) biopsy for thyroid nodules to check for cancerous cells.

Treatment :-

- Hypothyroidism : Replacement therapy with Synthetic thyroid hormone (Levothyroxine).
- Hyperthyroidism : Medications to reduce hormones production (such as methimazole or propylthiouracil), radioactive iodine therapy or surgery (thyroidectomy).
- Thyroid Nodules : observation, medications, or surgical removal depending on size and characteristics.
- Thyroiditis : Treatment varies based on the underlying causes (e.g. anti-inflammatory medications for thyroiditis).

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Management :-

- Regular Monitoring of thyroid functions through blood test.
- Medications adherence for thyroid hormones replacement or anti-thyroid drugs.
- Lifestyle modification (diet, exercise) to support thyroid health.
- Follow up with endocrinologist or thyroid Specialist as needed.

③ Disorders of Sex hormones :-

Disorders of sex hormones refer to conditions where there is an imbalance or dysfunctions in the production, regulation or action of sex hormones, primarily estrogen, progesterone, and testosterone.

Types of Disorders :-

1. Hypogonadism :-

- Condition where the sex glands (ovaries in females and testes in male) produce insufficient amount of sex hormones.
- Can be Congenital (present at birth) or acquired later in life.
- Causes include genetic disorders, autoimmune conditions, infections, or trauma.

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2. Polycystic Ovary Syndrome (PCOS):-

- Common hormonal disorder among women of reproductive age.
- Characterized by Irregular Menstrual Cycles, excess androgen (male hormones), and polycystic ovaries.
- Symptoms include Infertility, acne, hirsutism (excessive hair growth) and obesity.
- Linked to insulin ~~an~~ resistance and Metabolic Syndrome.

2. Endometriosis:-

- Conditions where tissue similar to the lining of the uterus grows outside the uterus.
- Causes Chronic pelvic pain, painful menstruation and infertility.
- Hormonal imbalance, particularly estrogens dominance, play a role in its development and progression.

3. Gynecomastia:-

- Enlargement of breast tissue in males
- Often due to hormonal imbalances (e.g. decrease testosterone, increased estrogen)
- Can occur during puberty, with aging, or due to certain medications and medical conditions.

Diagnosis:-

Based on symptoms, physical examination, hormones level (blood test), imaging studies (ultrasound, MRI) and sometimes Genetic testing.

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Management:-

Treatment depends on the specific disorder but may include hormone replacement therapy (HRT), Medications to regulate Menstrual Cycles (e.g. birth control pills), lifestyle changes (e.g. diet, exercise) and Surgery in some cases.

★ Nervous System:-

The nervous system is a complex network and cells that transmit signals between different parts of the body.

1. Structure: It comprises two main parts.

- **Central Nervous System (CNS):** Include the brain and spinal cord, responsible for processing information and coordinating responses.
- **Peripheral Nervous System (PNS):** Consists of nerves that extend throughout the body, connecting the CNS to limbs and organs.

2. Functions:-

- **Sensory Input:** Receives information from sensory receptors that detect stimuli both internally and externally.
- **Integration:** processes and interprets sensory input and decides on appropriate responses.
- **Motor Output:** Responds to integrated stimuli by activating effector organs (muscles or glands) through motor neurons.

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3. Neurons: Basic functional units of the nervous system, specialized cells that transmit electrical signals.

• Parts of neurons:

• Cell body: Contains the nucleus and other organelles.

• Axon: Long fiber that transmits signals away from the cell body.

• Dendrites: Branch like extensions that receive signals from other neurons.

4. Types of neurons:

• Sensory Neurons: Transmit sensory information from receptor to the CNS.

• Motor Neurons: Transmit motor signals from the CNS to muscles and glands.

• Interneurons: Found in the CNS, process information between sensory and motor neurons.

5. Nerve Impulses: Electrical signals that travel along neurons, allowing communication within the nervous system.

6. Brain: Central organ of the nervous system, responsible for processing sensory information, thoughts, emotions, and controlling voluntary and involuntary actions.

7. Spinal Cord: Connects the brain to the PNS, responsible for reflex actions and transmitting information between the brain and peripheral nerves.

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① Epilepsy:

Epilepsy is a neurological disorder characterized by recurrent seizures, which are sudden bursts of electrical activity in the brain.

- Epilepsy is a chronic disorder of the brain characterized by recurrent seizures.

Seizures: occur due to abnormal electrical activity in the brain, leading to temporary changes in behavior, awareness or sensation.

Types of seizures:

- Generalized Seizures: Involve both hemispheres of the brain and typically affect consciousness.

- Example: Tonic-clonic (formerly grand mal), absence, myoclonic, atonic.

- Focal (partial) Seizures: Begin in one area of the brain and may spread to other parts.

- Simple Focal Seizures: No loss of consciousness.

- Complex Focal Seizures: Impair awareness or consciousness.

Causes of Epilepsy: (Unknown Cause) but can be due to,

- Brain Injury or trauma.

- Stroke

- Brain tumors

- Infections (e.g. Meningitis)

- Genetic factor

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Diagnosis :- Based on Medical history, Symptoms, neurological examination and test such as EEG (electroencephalogram) MRI (Magnetic resonance imaging).

Treatment :-

- Medications: Antiepileptic drugs to prevent Seizures.
- Surgery: For Cases resistant to Medication, Surgery May be an option to remove the Seizure focus.
- Lifestyle Management: Avoiding triggers, Maintaining a regular Sleep Schedule and Managing Stress.

② Parkinson's disease :-

Parkinson's disease is a chronic and progressive Movement disorder characterized by tremors, stiffness, slowness of movement (bradykinesia) and impaired balance and Coordination.

Causes :-

Generally idiopathic (unknown cause), but thought to involve a combination of genetic and environment factors.

It involves the loss of dopamine-producing neurons in a specific part of the brain called the substantia nigra.

Symptoms :-

• Motor Symptoms :-

- Tremor (typically at rest) - Bradykinesia (Slowness of movement)
- Rigidity (Muscle stiffness) - Postural instability (balance problem)

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• Non-Motor Symptoms :-

- Cognitive changes (e.g. dementia)
- Sleep disturbances
- Mood disorder (e.g. depression)
- Autonomic dysfunction (e.g. Constipation, Urinary problem)

Diagnosis :- Based on medical history, neurological examination and assessment of symptoms. There's no specific diagnostic test: diagnosis is often clinical.

Treatment :-

- Medications: Dopamine replacement therapy (e.g. Levodopa) to alleviate symptoms by replenishing dopamine in the brain.
- Deep Brain Stimulation: Surgical procedure that can help manage symptoms in some cases.
- Physical Therapy: Helps maintain mobility and improve muscle tone.
- Lifestyle Changes: Exercise, a balanced diet and speech therapy may be beneficial.

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③ Stroke :-

A stroke occurs when blood flow to part of the brain is interrupted or reduced, depriving brain tissue of oxygen and nutrients.

Types:-

- **Ischemic stroke**: Caused by a blockage (usually a blood clot) in a blood vessel supplying the brain.
- **Hemorrhagic stroke**: Caused by a blood vessel that bursts and bleeds into the brain.

Symptoms:-

- Face drooping
- Arm weakness
- Speech difficulty
- Time to call emergency services

Risk factors:-

- High blood pressure
- High cholesterol
- Smoking
- Diabetes
- Age (risk increases with age)
- Family history

Treatment:-

- Immediate medical attention is crucial
- **Ischemic stroke**: Treatment with clot-busting drugs (Thrombolytics) or Mechanical clot removal (Thrombectomy).
- **Hemorrhagic stroke**: Focus on controlling bleeding and reducing pressure in the brain.

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Prevention :-

- Manage blood pressure and cholesterol levels.
- quit Smoking
- Control diabetes
- Maintain a healthy diet and exercise regularly.

★ psychiatric disorder :-

Psychiatric disorders are Conditions that affect mood, thinking and behavior, Causing distress and impairing functioning.

① Depression :-

Depression is a mood disorder characterized by persistent sadness, loss of interest or pleasure in activities.

Symptoms :-

- persistent Sadness, emptiness, or hopelessness
- Loss of interest or pleasure in most activities.
- Change in appetite or weight.
- Sleep disturbance (Insomnia or oversleeping)
- Fatigue or loss of energy
- Thoughts of death or suicide.
- Difficulty Concentrating or Making decisions
- Feeling of worthlessness or excessive guilt.

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Types:-

- **Major Depressive Disorder**: Characterized by severe symptoms that interfere with daily life for at least two weeks.
- **persistent Depressive disorder**: Chronic low-grade depression lasting for two years or more, with less severe symptoms.

Causes:-

- **Biological factors**: Imbalances in brain chemicals (neurotransmitters) like Serotonin, norepinephrine and dopamine.
- **Genetic Factors**: family history of depression increase the risk.
- **Psychologic factors**: Stressful life events, trauma, or chronic illness.

Treatment:-

- **Psychotherapy**: Cognitive-behavioral therapy (CBT), interpersonal therapy (IPT) or psychodynamic therapy to address negative thought patterns and behaviors.
- **Medications**: Antidepressants such as SSRIs (selective Serotonin Reuptake inhibitor) or SNRIs (serotonin-norepinephrine Reuptake inhibitor) to regulate brain chemistry.
- **Lifestyle Changes**: Regular exercise, healthy diet, adequate sleep, and reducing stress.
- **Support network**: Family support, support groups, or counseling.

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② Schizophrenia:-

Schizophrenia is a chronic and severe mental disorder characterized by disturbances in thinking, emotions, perceptions and behaviors.

Symptoms:-

- Positive Symptoms: Hallucination (seeing or hearing things that aren't there) delusions (false beliefs), disorganized thinking and speech.
- Negative Symptoms: Lack of Motivation, reduced emotion expression, social withdrawal and difficulty in functions.

Causes:-

- Biological factors: Genetic predisposition, abnormal brain structure or chemistry.
- Environmental Factor: prenatal exposure to viruses, stress during early development and psychosocial stress.

Treatment:-

- Antipsychotic Medications: Help manage symptoms by affecting neurotransmitters in the brain.
- Psychosocial Intervention: Therapy, family education and support, vocational rehabilitation and social skill training to improve functioning.

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③ Alzheimer's disease:-

Alzheimer's disease is a progressive neurological disorder that causes gradual cognitive decline and memory loss.

Symptoms:-

- Early Stage: Forgetfulness, difficulty remembering recent events or conversations.
- Middle Stage: Confusion, disorientation, trouble with language and communication.
- Late Stage: Severe memory loss, inability to recognize family and friends, difficulty with basic tasks.

Causes:-

- Build-up of plaques (amyloid-beta protein) and tangles (tau protein) in the brain.
- Loss of connections between nerve cells (neurons) disrupts communication.

Diagnosis:-

- Clinical evaluation, including cognitive tests and medical history.
- Brain imaging (MRI, CT) to detect changes in brain structure.

Treatment:-

- No cure, but medication (e.g. cholinesterase inhibitor) can temporarily improve symptoms or slow progression.

Non-drug therapies: Cognitive stimulation, occupational therapy, and caregiver support.

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* Gastrointestinal System:-

The gastrointestinal (GI) System, also known as digestive system, is a complex network of organs and processes that work together to process food, extract nutrients and eliminate waste from the body.

- It includes the following main organs:

- Mouth
- Small intestine
- Esophagus
- Large intestine
- Stomach
- Liver, Gall bladder and pancreas.

① Peptic Ulcer:-

peptic ulcers are sores that develop on the lining of the stomach, small intestine or esophagus.

Cause:-

◦ Helicobacter pylori (H. pylori) infection:-

This bacterium weakens the protective mucous coating of the stomach and duodenum, allowing acid to damage the lining.

◦ Nonsteroidal anti-inflammatory drugs (NSAIDs):-

Regular use of medications like aspirin, ibuprofen and naproxen can irritate the stomach lining and lead to ulcers.

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Symptoms:-

- Burning Stomach pain: Usually occurs between Meals and in the early morning.
- Nausea, Vomiting, and bloating.
- Loss of appetite and unexplained weight loss

Diagnosis:-

- Endoscopy: A thin, flexible tube with a camera (endoscope) is passed down the throat to examine the stomach and duodenum.
- Test for H. pylori: Blood, stool, or breath tests can detect antibodies to the bacterium or its byproducts.

Treatment:-

- Antibiotics: Use to eradicate H. pylori infection.
- proton pump inhibitor and H₂-receptor antagonists:- Reduce stomach acid production to promote healing.
- Antacids: provide temporary relief by neutralizing stomach acid.