UNIT-4

SECTION-A

1.What is proteinuria?

When healthy kidneys filter fluid, minerals and wastes from the blood, they usually do not allow large amounts of serum protein to escape into the urine. But when kidneys aren't filtering properly, proteinuria can occur, meaning that an abnormal amount of protein is present in the urine.

2.What is gout?

Gout is a kind of arthritis that causes sudden pain and swelling in joints. It usually shows up first in the big toe, but it can also occur in other joints. Gout usually happens when uric acid builds up in the blood (a condition called hyperuricemia).

3. What are plasma proteins?

Plasma proteins are proteins found in the blood plasma, the clear, protein-rich fluid which is left behind when platelets, red blood cells, and white blood cells are removed from the blood.

4.W hat is multiple myeloma?

Multiple myeloma, also known as Kahler's disease, is a type of blood cancer. There's no cure, but treatments can slow its spread and sometimes make symptoms go away.

5.What are warning signs of multiple myeloma?

The most common signs and symptoms of multiple myeloma are Fatigue. Bone problems. Kidney problems. Low blood counts. Frequent infections.

6.What is albinism?

Albinism is a rare group of genetic disorders that cause the skin, hair, or eyes to have little or no color. Albinism is also associated with vision problems.

SECTION-B

1.Explain the origin of plasma proteins.

In the embryo,the primitive plasma and the plasma proteins are produced either by secretion or actual solution of the mesenchymal cells.The albuminfractionis the first to be formed.

In the adults all the four fractions are produced by the liver.Fibrinogen,prothrombin and albumin are manufactured only in the liver.

a)From the disintegrated blood cells,

b)From the reticulo-endothelial system

c)From the tissue cells in general

d)From lymphoid nodules.

2.Explain the functions of plasma proteins.

- Fibrinogen and prothrombin are essential for coagulation of blood.

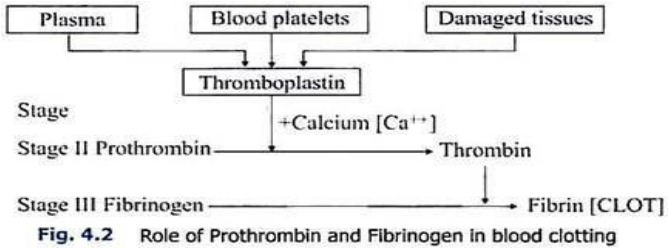


Fig. 4.2 Role of Prothrombin and Fibrinogen in blood clotting

- All the three proteins take part.Albumin having the smallest and the most symmetrical molecule exerts the maximum osmotic pressure.Osmotic pressure depends upon a number of molecules in the solution.
- The plasma proteins exert a great influence upon the suspension stability of blood.This is dependent on fibrinogen,less on globulin and least on albumin.
- They act as buffers in maintaining acid-base balance.
- They serve as a storehouse of proteins.
- Help co carriage by forming carbamino proteins
- They combine with certain substances and help to carry them in the blood stream.

3.Explain about the fanconi syndrome.

- Fanconi syndrome (FS) is a rare disorder that affects the filtering tubes (proximal tubules) of the kidney.
- Normally, the proximal tubules reabsorb the minerals and nutrient into the bloodstream that are necessary for proper functioning.
- In FS, the proximal tubules instead release large amounts of these essential metabolites into the urine. These essential substances are include water,glucose,phosphate,bicarbonates,uric acid etc.
- The resulting lack of essential metabolites can cause dehydration, bone deformities, and a failure to thrive.There are treatments available that can slow or stop FS progression.

Symptoms :Thrist,urination,vomiting,slow growth,rickets etc.

Causes :

- Cystinosis is the most common cause of FS. It's a rare inherited disease. In cystinosis, the amino acid cystine accumulates throughout the body.

Treatment :

- For children with inherited FS, the first line of treatment is to replace the essential substances that are being eliminated in excess by the damaged kidneys.
- Replacement of these substances may be by mouth or by infusion.

4.What is Wilson's disease?

- Wilson's disease, also known as hepatolenticular degeneration and progressive lenticular degeneration, is a rare genetic disorder that causes copper poisoning in the body.
- In a healthy body, the liver filters out excess copper and releases it through urine.
- With Wilson's disease, the liver cannot remove the extra copper properly. The extra copper then builds up in organs such as the brain, liver, and eyes.
- Early diagnosis is crucial for stopping the progression of Wilson's disease. Treatment may involve taking medication or getting a liver transplant.

- Delaying or not receiving treatment can cause liver failure, brain damage, or other life-threatening conditions.

- **Symptoms:**
- The signs and symptoms of Wilson's disease vary widely, depending on which organ is affected.
- Many of these symptoms, such as jaundice and edema, are the same for other conditions like liver and kidney failure.

Diagnosis:

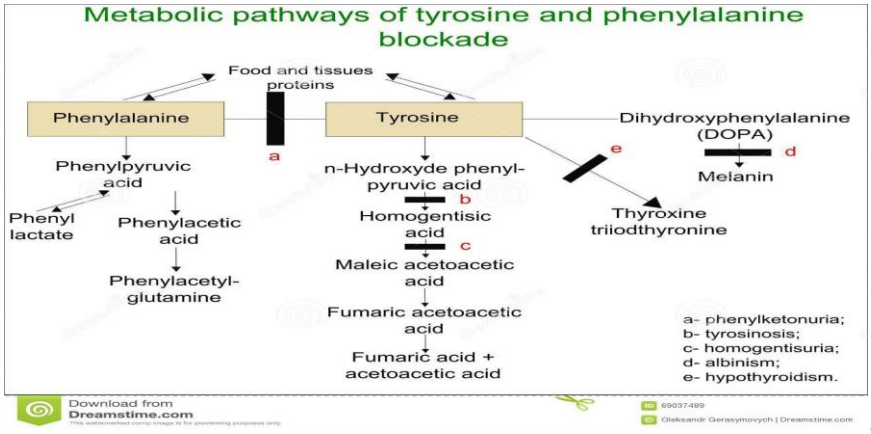
- Successful treatment of Wilson's disease depends upon timing more than medication. Treatment often happens in three stages and should last a lifetime. If a person stops taking the medications, copper can build back up again.

SECTION-C

1. Explain about the following disorders.a).Albinism b)Tyrosinosis c)Phenyl ketonuria d)Alkaptonuria e)MSUD

Albinism:

- This condition appears in the total absence of tyrosinase inside the melanocytes in the skin.
- The black pigment melanin is not formed in the skin, eyes and hair.
- This inherited condition occurs to a greater or less extent in all types of organism.
- The diagnostic advice is for the prevention of exposure of sunlight and protection of the eyes by wearing dark glasses.



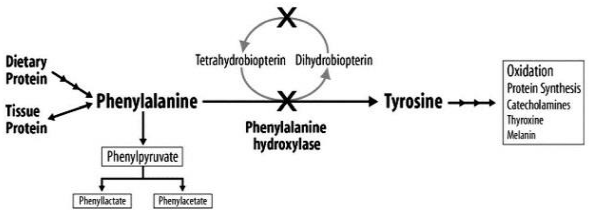
Tyrosinosis:

- This syndrome is due to the absence either of hepatic p-hydroxy phenylpyruvate hydroxylase.
- The patient excretes large quantities of tyrosine in the urine.
- Diet rich in tyrosine causes the excretion of other p-hydroxyphenyl acids
- Including 3,4-dihydroxyphenylalanine and p-hydroxyphenyl lactic acid.

Phenylketonuria:

- This inherited disorder appears in the absence of phenylalanine hydroxylase which is responsible for the conversion of phenylalanine to tyrosine.
- As a result,alternative catabolizes of phenylalanine are produced.These include phenyl pyruvic acid,deamination product of phenylalanine,phenyl aceticacid,the decarboxylation and oxidation product of phenyl pyruvic acid.
- Much of the phenyl-acetyl-glutamine is excreted in the urine.
- Mental retardation develops among infants and children.
- Patients with pku tend to have a deficiencyof serotonin.

Phenylketonuria (PKU)



- The accumulation of phenylalanine also impairs melanin synthesis and children with this defect tend have fair skin and fair hair.
- Excess of phenylalanine in the blood leads to excretion of the aminoacid into the intestine.
- Early diagnosis and extreme restriction of phenylalanine intake is effective in preventing this disorder.

Alkaptonuria:

- This condition is characterized by the excretion of homogentisic acid in the urine owing to the lack of homogentisic acid oxidase.
- This condition is often found in infancy.
- Mental retardation develops among infants and children.
- The clinical manifestation is dark urine due to the oxidation of homogentisic acid in air.
- In this disorder,several grams of homogentisic acid are excreted daily.
- This condition is present at birth and persists throughout life.

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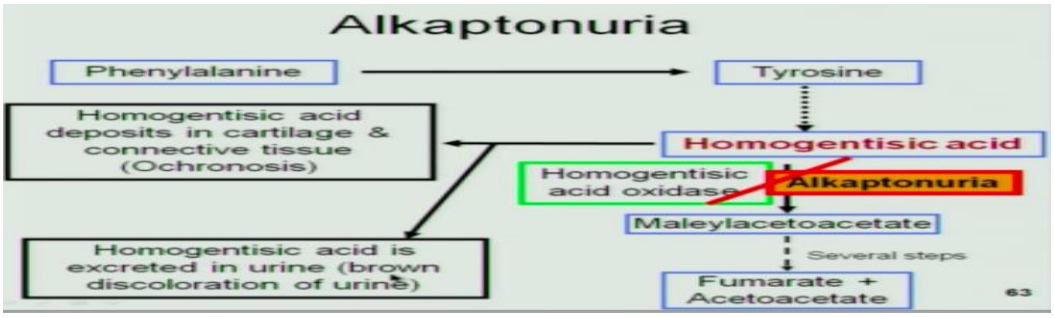


Maple syrup urine disease:

- This syndrome is characterized by the absence of the enzymes required for the oxidative decarboxylation of the keto acids derived from the branched chain amino acids-valine,leucine and isoleucine.
- The urinary excretion of these keto acids produce an odour like that of maple syrup or of burnt sugar.
- This disorder is recognized by central nervous system manifestations of flaccidity and apnea.
- The infant is difficult to feed and may vomit.
- Death may occur by the end of the first year life without treatment

3.Explain about the hartnup's disease,cystinuria,Homocystonuria.

- Hartnup's disease:
- It is a hereditary disease characterized by a pellagra-like rash and mental deterioration in the abnormal metabolism of tryptophan.
- The urine of the patients contain significantly increased amounts of indole acetic acid as well as tryptopan
- The urinary excretion comes to normal after administration of broad spectrum.

Cystinuria:

- It is an inherited metabolic disease in which lysine,arginine,ornithine,and cystine are excreted in the urine in large amounts.
- It is due to renal transport defect.
- It is a misnomer,so that cystinelysinuria may be preferred as the descriptive term for this disease.
- It is an insoluble amino acid which may precipitate in the kidney tubules to form cystine calculi in cystinuric patients.

Homocystonuria:

- In this condition, homocystine together withs-adenosylmethionine is excreted in the urine and plasma methionine levels are elevated.
- The clinical findings of this disease are the occurrence thrombosis,osteoporosis,dislocated lenses in the eyes and frequently mental retardation.
- This condition appears due to the lack of cystathionine synthetase in the liver due to which both homocystine and methionine are accumulated in blood and urine.
- Alow methionine and ahigh cystine diet prevent this condition if treated earlier.

3..E xplain about the proteinuria.

- People with proteinuria have unusually high amounts of protein in their urine. The condition is often a sign of kidney disease.
- kidneys are filters that don’t usually let a lot of protein pass through.

Symptoms:shortness of breath,fatigue,loss of appetite,etc.

Causes:dehydration,inflammation,fever,stress,kidney stones etc.

- The two most common are diabetes and high blood pressure.
- Obesity
- Age over 65
- A family history of kidney disease
- Islander.

Diagnosis:

- A urine test called a urinalysis .A lab technician will dip in a stick in to a sample with chemicals on the end. If the stick changes color, it's a sign of too much protein..

UNIT-5

SECTION-A

1.What is Niemann-pick disease?

Niemann-Pick disease is type of lysosomal storage disorder. Types A and B are sphingolipidoses and are caused by a buildup of sphingomyelin in the tissues.

Type C is a lipidosis that is caused by a build up of cholesterol and other fats (lipids) in the cells. This disease causes many neurologic problems.

2.What is Gaucher's disease?

Abnormal amounts of cerebrosides accumulate in the liver, spleen, bone marrow, and lymph nodes. The defective enzyme is glucocerebrosidase.

3.What are signs and symptoms of atherosclerosis?

Common symptoms include: chest pain or angina. pain in your leg, arm, and anywhere else that has a blocked artery. shortness of breath. fatigue. confusion, which occurs if the blockage affects circulation to the brain.

4.What is Atherosclerosis?

Atherosclerosis is a disease of the arteries characterized by the deposition of fatty material on their inner walls.

5.How do lipid storage disorders affect the body?

Lipid storage disorders cause an unhealthy excess of lipids in cells of the body. Often these build up in important tissues, like those of the liver, spleen, and brain. These stores of lipids eventually interfere with the proper functioning of the tissues and cells and cause irreversible damage.

6.How hyperlipoproteinemia is diagnosed?

A doctor can diagnose hyperlipoproteinemia with a blood test. Sometimes, family history is useful.

Other diagnostic tests might measure thyroid function, glucose, protein in the urine, liver function, and uric acid

7.What is a lipid disorder?

This disorder have high blood levels of low-density lipoprotein (LDL) cholesterol, and fats called triglycerides, or both. High levels of these substances increase risk for developing heart disease.

SECTION-B

1.Write a note on Atherosclerosis.

Atherosclerosis is a disease of the arteries characterized by the deposition of fatty material on their inner walls.

Signs and Symptoms:

- Arrhythmia, an unusual heartbeat
- Pain or pressure in upper body, including your chest, arms, neck, or jaw. This is known as angina.
- Shortness of breat
- Numbness or weakness in your arms or leg
- A hard time speaking or understanding someone who's talking
- Drooping facial muscle
- Paralysis
- High blood pressure
- Kidney failure

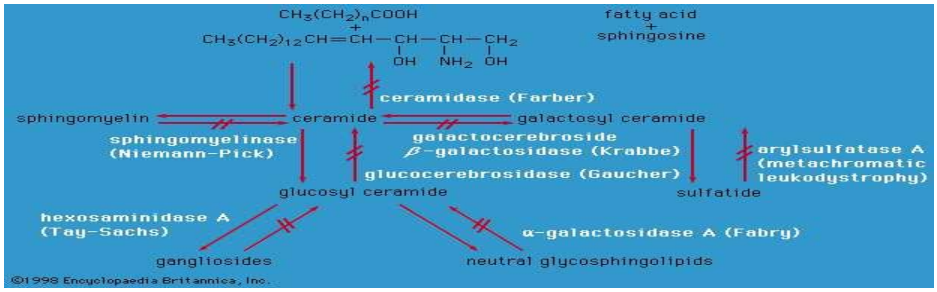
Diagnosis

- Angiogram, in which your doctor puts dye into your arteries so they'll be visible on an X-ray
- Ankle-brachial index, a test to compare blood pressures in your lower leg and arm

- Blood tests to look for things that raise your risk of having atherosclerosis, like high cholesterol or blood sugar
- Chest X-ray to check for signs of heart failure
- CT scan or magnetic resonance angiography (MRA) to look for hardened or narrowed arteries
- EKG, a record of your heart's electrical activity

2.Discuss about the disorders of lipid metabolism.

- Lipids are large, water-insoluble molecules that have a variety of biological functions, including storing energy and serving as components of cellular membranes and lipoproteins.
- Cells that line the small intestine absorb dietary lipids and process them into lipoprotein particles that enter the circulation via the lymphatic system for eventual uptake by the liver.
- Triglycerides, cholesterol, and fat-soluble vitamins are transported through the blood by these lipoprotein particles.



- Lipid storage disease, any of a group of relatively rare hereditary disorders of fat metabolism, characterized by the accumulation of distinctive types of lipids, notably cerebrosides, gangliosides, or

sphingomyelins, in various body structures.

Disease	Enzyme Deficiency	Lipid Accumulating[1]	Clinical Symptoms
Tay-Sachs disease	Hexosaminidase A	Cer—Glc—Gal(NeuAc)÷GalNAc G_{M2} Ganglioside	Mental retardation, blindness, muscular weakness.
Fabry's disease	α-Galactosidase	Cer—Glc—Gal÷Gal Globotriaosylceramide	Skin rash, kidney failure (full symptoms only in males; X-linked recessive).
Metachromatic leukodystrophy	Arylsulfatase A	Cer—Gal÷OSO_3 3-Sulfogalactosylceramide	Mental retardation and psychologic disturbances in adults; demyelination.
Krabbe's disease	β-Galactosidase	Cer÷Gal Galactosylceramide	Mental retardation; myelin almost absent.
Gaucher's disease	β-Glucosidase	Cer÷Glc Glucosylceramide	Enlarged liver and spleen, erosion of long bones, mental retardation in infants.
Niemann-Pick disease	Sphingomyelinase	Cer÷P—choline Sphingomyelin	Enlarged liver and spleen, mental retardation; fatal in early life.
Farber's disease	Ceramidase	Acyl÷Sphingosine Ceramide	Hoarseness, dermatitis, skeletal deformation, mental retardation; fatal in early life.

[1]NeuAc, *N*-acetylneuraminic acid; Cer, ceramide; Glc, glucose; Gal, galactose. ÷, site of deficient enzyme reaction.

2.Write a note on Hypolipoproteinemia.

Definition:

Hypolipoproteinemia (or hypolipidemia) is the lack of fat in the blood.

Description:

Although quite rare, hypolipoproteinemia is a serious condition. Blood absorbs fat from food in the intestine and transports it as a combined package with proteins and other chemicals like cholesterol. Much of the fat goes straight into the liver for processing

The cholesterol, a waste product, ends up in the bile. The proteins act as vessels, carrying the other chemicals around. These packages of fat, cholesterol, and proteins are called lipoproteins.

Causes and symptoms:

Low blood fats can be the result of several diseases, or they can be a primary genetic disease with other associated abnormalities.

- Malnutrition is a lack of food, including fats, in the diet.

- Malabsorption is the inability of the bowel to absorb food, causing malnutrition.
- Anemia and hyperthyroidism also reduce blood fats.
- Rare genetic conditions called hypobetalipoproteinemia and abetalipoproteinemia cause malabsorption plus nerve, eye, and skin problems in early childhood.
- Tangier disease, causes only the cholesterol to be low. It also produces nerve and eye problems in children.

Symptoms are associated more closely with the cause rather than the actual low blood fats.

Diagnosis:

Blood studies of the various fat particles help identify both the low and high fat diseases. These tests are often done after an overnight fast to prevent interference from fat just being absorbed from food. Fats and proteins are grouped together and described by density—high-density lipoproteins (HDL), low-density lipoproteins (LDL), and very low-density lipoproteins (VLDL). There are also much bigger particles called chylomicrons.

Treatment

Supplemental vitamin E helps children with the betalipoprotein deficiencies. There is no known treatment for Tangier disease. Treatment of the causes of the other forms of low blood fats reverses the condition.

3.Write a short note on a)gaucher's disease b)Niemann-pick disease

a)Gaucher's disease:

- Abnormal amounts of cerebrosidesaccumulate in the liver, spleen, bone marrow, and lymph nodes. The defective enzyme is glucocerebrosidase.
- Two distinctive syndromes: (1) An acute cerebral form chiefly affects infants, who appear normal at birth but soon become apathetic and retarded in their development; enlargement of their abdomen is followed by severe nervous system symptoms, and death usually occurs during the first year of life.
- (2) A more chronic form that may become evident at any age is characterized by an enlargement of the spleen, by anemia, and by a patchy brown pigmentation of the skin; the bones show characteristic changes in shape.
- Most people who have Gaucher disease have varying degrees of the following problems:
- Abdominal complaints. Because the liver and especially the spleen can enlarge dramatically, the abdomen can become painfully distended.
- **Skeletal abnormalities.** Gaucher disease can weaken bone, increasing the risk of painful fractures. It can also interfere with the blood supply to your bones, which can cause portions of the bone to die.
- **Blood disorders.** A decrease in healthy red blood cells (anemia) can result in severe fatigue. Gaucher disease also affects the cells responsible for clotting, which can cause easy bruising and nosebleeds.
- Gaucher disease is passed along in an inheritance pattern called autosomal recessive. Both parents must be carriers of a Gaucher changed (mutated) gene for their child to inherit the condition.

b)Niemann-pick disease:

- Niemann-Pick disease**,** inherited metabolic disorder in which a deficiency of the enzyme sphingomyelinase impairs the breakdown of the phospholipids lecithin and sphingomyelin, causing them to accumulate in various body tissues
- Symptoms consist of extreme liver and spleen enlargement, mental retardation, and a brownish-yellow skin discoloration; foamy cells containing phospholipids are found in several organs.
- There are five distinct varieties of the disease, the most common of which is the acute infantile form (type A). Affected infants are retarded in growth; they lose weight and undergo a decline in mental and neurologic functions, usually dying by age four. In the chronic visceral form (type B), development is normal for several years until poor muscle coordination and liver enlargement become apparent.
- Type C, which appears in adolescence, is similar to the acute infantile form, as is type D, which is found among a small group of people whose common ancestry is traced to Nova Scotia..

SECTION-C

1.Explain about the hyperlipoproteinemia.

Hyperlipoproteinemia is a common disorder. It results from an inability to break down lipids or fats in your body, specifically cholesterol and triglycerides.

The type depends on the concentration of lipids and which are affected.

High levels of cholesterol or triglycerides are serious because they’re associated with heart problems.

Causes of hyperlipoproteinemia:

Primary hyperlipoproteinemia is often genetic. It's a result of a defect or mutation in lipoproteins.

Secondary hyperlipoproteinemia is the result of other health conditions that lead to high levels of lipids in the body. These include:

- diabetes
- hypothyroidism
- pancreatitis
- use of certain drugs, such as contraceptives and steroids
- certain lifestyle choices

Types of primary hyperlipoproteinemia:

There are five types of primary hyperlipoproteinemia:

Type 1 is an inherited condition. It causes the normal breakdown of fats in the body to be disrupted. A large amount of fat builds up in blood as a result.

Type 2 runs in families. It's characterized by an increase of circulating cholesterol, either low-density lipoproteins (LDL) alone or with very-low-density lipoproteins (VLDL). These are considered the "bad cholesterols."

Type 3 is a recessively inherited disorder in which intermediate-density lipoproteins (IDL) accumulate in the blood. IDL has a cholesterol-to-triglycerides ratio that's higher than that for VLDL.

Type 4 is a dominantly inherited disorder. It's characterized by high triglycerides contained in VLDL. The levels of cholesterol and phospholipids in your blood usually remain within normal limits.

Type 5 runs in families. It involves high levels of LDL alone or together with VLDL.

Symptoms :

Lipid deposits are the main symptom of hyperlipoproteinemia. The location of lipid deposits can help to determine the type. Some lipid deposits, called xanthomas, are yellow and crusty.

2. Explain about the Hypolipoproteinemia.

Definition:

Hypolipoproteinemia (or hypolipidemia) is the lack of fat in the blood.

Description

Hypolipoproteinemia is a serious condition. Blood absorbs fat from food in the intestine and transports it as a combined package with proteins and other chemicals like cholesterol. Much of the fat goes straight into the liver for processing.

The cholesterol, a waste product, ends up in the bile. The proteins act as vessels, carrying the other chemicals around. These packages of fat, cholesterol, and proteins are called lipoproteins.

Causes and symptoms

Low blood fats can be the result of several diseases, or they can be a primary genetic disease with other associated abnormalities.

- Malnutrition is a lack of food, including fats, in the diet.
- Malabsorption is the inability of the bowel to absorb food, causing malnutrition.
- Anemia and hyperthyroidism also reduce blood fats.

- Rare genetic conditions called hypobetalipoproteinemia and abetalipoproteinemia cause malabsorption plus nerve, eye, and skin problems in early childhood.
- Tangier disease, causes only the cholesterol to be low. It also produces nerve and eye problems in children.

Symptoms are associated more closely with the cause rather than the actual low blood fats.

Diagnosis:

Blood studies of the various fat particles help identify both the low and high fat diseases.. Fats and proteins are grouped together and described by density—high-density lipoproteins (HDL), low-density lipoproteins (LDL), and very low-density lipoproteins (VLDL). There are also much bigger particles called chylomicrons.

Treatment:

Supplemental vitamin E helps children with the betalipoprotein deficiencies. There is no known treatment for Tangier disease. Treatment of the causes of the other forms of low blood fats reverses the condition.

3. What is a lipid disorder?Explain it.

This disorder have high blood levels of low-density lipoprotein (LDL) cholesterol, and fats called triglycerides, or both. High levels of these substances increase your risk for developing heart disease.

Cholesterol

Two major forms of cholesterol :

low-density lipoprotein (LDL) and high-density lipoprotein (HDL).

LDL, sometimes known as “bad cholesterol,” is made by a body and also absorbed by body from cholesterol-rich foods such as red meat and dairy products. LDL can combine with other fats and substances in theblood, creating blockages in arteries.

Blockages in arteries can reduce blood flow and cause serious health problems such as heart disease, heart attack, or stroke. Because of its potential effects, doctors recommend lower levels of LDL.

HDL, sometimes known as “good cholesterol,” has a protective effect on heart.

Triglycerides

A triglyceride is a type of fat to get mostly from the food to eat. Body also produces it when it converts excess calories to fat for storage. Some triglycerides are necessary for certain cell functions, but too much is unhealthy.

Foods high in certain types of fats, certain medical conditions, and other factors can cause high blood cholesterol and high triglycerides.

Food

Two types of fat are known to increase cholesterol levels.

Saturated fat: Saturated fats can increase your LDL levels. Some plant-based foods, such as palm oil and coconut oil, contain saturated

- fats.
- milk
- butter
- steak

Trans fats: Trans-fatty acids, are worse than saturated fats because they can raise your LDL levels and lower your HDL levels. Some trans fats are found naturally in animal products.

Medical conditions

Certain medical conditions can affect your cholesterol levels

- diabetes
- hypothyroidism
- metabolic syndrome
- Cushing's syndrome
- polycystic ovary syndrome (PCOS)
- kidney disease

Symptoms :

High cholesterol typically doesn't cause any symptoms. Symptoms may only appear after the increased cholesterol has caused significant damage.