1. **Thalassemia**

The thalassemia is a blood disorder passed through the families (inherited) in which the body makes an abnormal form of Hb.

Definition: It is an autosomal recessive genetic disorder that results inadequate production of normal Hb.

Etiology: Caused by genetic mutation, or a deletion of certain key gene fragments.

Family history of thalassemia

It is most common in people from Asia, the middle east, Africa, and Mediterranean countries such as Greece and Turkey.

Types of thalassemia:

1. Alpha thalassemia: It is a most common type of thalassemia. Here alpha globin protein chains are changed or absent or reduced.
2. Beta thalassemia: here beta globin chains are absent or reduced.
3. Thalassemia major: thalassemia major is the severe form of beta thalassemia. It develops when beta globin genes are missing.
4. Thalassemia minor: in alpha minor cases, two genes are missing. In beta minor, one gene is missing. People with thalassemia minor do not usually have any symptoms.

Clinical manifestations:

* Individual with alpha thalassemia may have mild anemia and typically asymptomatic.
* Children born with beta- thalassemia major (Cooley anemia) are normal at birth but develop severe anaemia during first year of life.
* Individual with thalassemia major is diagnosed early in life because the lack of Hb becomes quickly apparent.
* Children also have pain, failure to thrive, frequent infections, diarrhoea, splenomegaly, hepatomegaly, jaundice from RBC hemolysis, and bone marrow hyperplasia.
* Other symptoms include bone deformities is face
* Fatigue
* Growth failure
* Shortness of breath
* Jaundice

Diagnostic evaluation:

* History collection
* Physical examination
* Blood investigations
* Amniocentesis (Fetal diagnosis for specific type of thalassemia.

Management:

Medical management: blood transfusion

Client should not take iron supplements because it causes high amount of iron to build up in body which can be harmful.

Genetic counselling and testing for families should be encouraged.

Surgical management:

Bone marrow transplantation

Splenectomy

Nursing management:

1. **Polycythaemia**

People with polycythaemia have an increase in haematocrit, haemoglobin, or red blood cell count above the normal limits.

Definition: It is an abnormally increased concentration of haemoglobin in the blood, either through reduction of plasma volume or increase in red cell numbers.

Risk factors:

* Chronic hypoxia
* Cigarette smoking
* Familial and genetic predisposition

Types:

1. Primary polycythaemia: Primary polycythaemia occurs when excess red blood cells are produced because of an abnormality of the bone marrow. Often excess white blood cells and platelets are also produced.
2. Secondary polycythaemia: It is usually due to increased erythropoietin production either in response to chronic hypoxia or form an erythropoietin secreting tumour.
3. Relative polycythaemia: It is an increase in RBC numbers without an increase in total RBC mass. This is caused by loss of plasma volume with resultant hemo-concentration, as seen in severe dehydration related to vomiting and diarrhoea.
4. Stress polycythaemia: It is a term applied to a chronic state of low plasma volume which is seen commonly in active, hardworking, anxious, middle-aged men. In these people, the red blood cell volume is normal, but the plasma volume is low.

Clinical manifestations:

Symptoms results from increased blood volume:

* Cyanosis
* Reddened face with engorged retinal veins
* Itching after bath
* Feeling of fullness in head with headache
* Weakness, fatigue, dizziness
* Tinnitus (ringing or buzzing in the ears)
* Paraesthesia, numbness, burning or weakness in hands and legs
* Visual disturbances
* Nose bleeding
* Abdominal bloating

Symptoms from increased viscosity:

* Angina
* Dyspnoea
* Hypoxia
* Bone and joint pain
* Thrombophlebitis

Other symptoms are

* Weight loss
* Breathing difficulty when lying down
* Chronic cough
* Night sweats, sleep disturbances
* Burning sensations over fingers or toes
* Splenomegaly and hepatomegaly
* Formation of blood clots in the blood vessels

Diagnostic evaluation:

* History collection and physical examination
* Blood tests
* Bone marrow aspiration or biopsy
* Chest X-ray
* Electrocardiogram
* Echocardiogram
* Sp02 monitoring

Management:

Medical management:

* Phlebotomy: Drawing a certain amount of blood out of the veins in a procedure called as phlebotomy. It is the first treatment option for people with polycythaemia.

This reduces the number of blood cells and decreases blood volume, making it easier for blood to function properly.

* Low dose of aspirin: to prevent blood clots.
* Interferon alpha may be used to stimulate the immune system to fight the over production of red blood cells.
* Hydroxyurea is a drug used to suppress the bone marrow’s ability to produce blood cells may be used.
* Antihistamines to reduce the itching
* Lifestyle and home remedies are as follows
* Exercise
* Avoid tobacco
* Avoid extreme temperatures

Nursing diagnosis:

Impaired tissue perfusion related to phlebotomy as evidenced by cyanosis

Acute pain related to surgical intervention as evidenced by verbalization

Impaired breath pattern related to decreased level of RBC in blood as evidenced by dyspnea.

Imbalanced nutrition less than body requirement related to less oral intake of food as evidenced by weight loss.

Fear and anxiety related to outcome of disease condition as evidenced by frequent doubts.

Knowledge deficit regarding deficit regarding home care management as evidenced by frequent doubts.

**3. Bleeding disorders**

A group of disorder characterized by defective haemostasis with abnormal bleeding.

Etiology:

Due to vascular abnormality

Due to platelet abnormality

Disorder of coagulation factor

Combination of all these as in disseminated intravascular coagulation

Disorders of haemostasis

Classification:

1. Disorders of blood vessels
* Scurvy
* Senile purpura
1. Disorders of coagulation
* Extrinsic
* Intrinsic
* Combined
1. Disorders of platelets
* Thrombocytopenia, TIP,HUS,DIC
* Aspirin therapy, thrombasthenia
1. Other disorders
* Post transfusion purpura

Disorders of blood vessels:

Vitamin C deficiency leads to scurvy.

Disorders of coagulation:

Hemophilia A: It is a genetic deficiency in clotting factor V111, which causes it is inherited as an X -linked recessive trait, though there are cases which arise from spontaneous mutations.

Symptoms:

* Bruising
* Spontaneous bleeding
* Bleeding into joints and associated pain and swelling
* Gastrointestinal tract and urinary tract haemorrhage
* Blood in the urine or stool
* Prolonged bleeding from cuts, tooth extraction and surgery

Treatment:

* Infusion of cryoprecipitate or desmopressin acetate
* Desmopressin injection or STIMATE nasal spray

Hemophilia B (factor 1X deficiency) : It is a rare genetic bleeding disorder in which affected individuals have insufficient levels of a blood protein called factor 1X is a clotting factors.

Symptoms:

* Bruising
* Spontaneous bleeding
* Bleeding into joints and associated pain and swelling
* Gastrointestinal tract and urinary tract hemorrhage
* Blood in the urine or stool

Diagnosis:

 History collection, Lab test

Treatment:

Infusing the missing clotting factor

Von Willebrand disease: Hereditary deficiency or abnormality of the von Willebrand factor in the blood, a protein that affects platelet function.

Symptoms:

History of bleeding problem

Bleeds excessively after a skin cut, tooth extraction, tonsillectomy, or other surgery

Hormonal changes, stress, pregnancy, inflammation, and infection

Diagnostic evaluation:

* Normal platelet count
* Prolonged bleeding time
* Reduced von Willebrand factor level

Complications: after surgery haemorrhaging may occur. This condition is worsened using aspirin and other nonsteroidal anti-inflammatory drugs. Women may have risks during pregnancy and childbirth.

Disorders of platelets: Thrombocytopenia: It is any disorder in which the platelet count is below 1,50,000/cc of blood.

Causes:

Low production of platelets in the bone marrow (aplastic anemia, cancer in the bone marrow, infection in the bone marrow)

Increased breakdown of platelets in the blood stream(immune thrombocytopenic purpura, drug induced immune thrombocytopenia, drug induced nonimmune thrombocytopenia, disseminated intravascular coagulation (DIC), hypersplenism (an enlarged spleen)

Increased breakdown of platelets in the spleen or liver

Symptoms:

* Bruising
* Nose bleeds
* Rash

 Diagnostic evaluation:

* History collection
* Blood tests

Complications:

* Haemorrhage
* Gastrointestinal bleeding
* Intracranial haemorrhage

**Disseminated intravascular coagulation**: A condition affecting the blood’s la ability to clot and stop bleeding. In DIC, abnormal clumps of thickened blood clots form inside blood vessels. these abnormal clots use up the blood’s clotting factors, which can lead to massive bleeding in other places.

Causes include inflammation, infection, and cancer.

Risk factors: blood transfusion reaction

* Cancer
* Bacterial, fungal infection in the blood
* Liver disease
* Pregnancy complications (such as placenta that is left behind after delivery)
* Recent surgery or anaesthesia
* Severe tissue injury (burns and head injury)

Symptoms:

* Bleeding from many sites in the body
* Blood clots
* Bruising
* Drop in blood pressure
* Shortness of breath
* Confusion, memory loss changes of behaviour
* Fever

Diagnostic evaluation:

* History collection and physical examination
* Blood investigations

Treatment:

Plasma transfusion to replace blood clotting factors

Blood thinner medicine (heparin) to prevent blood clotting