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Early Clinical Exposure
A Case Based Approach in
Clinical Physiology

As per the Revised Competency-based Medical Education Curriculum (NMC-2024)



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2nd Edition

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Genuine?
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1 CHAPTER

Hematology

PY2.5 Describe anemias and jaundice. Discuss their physiological principles of management.

PY2.9 Describe hemostasis, coagulation pathways, mechanism of action of anticoagulants and briefly discuss pathophysiological aspects of bleeding and clotting disorders (e.g., hemophilia, purpura).

LEARNING OBJECTIVES

After finishing this module, you should be able to:

- Analyze the clinical problem on the basis of its signs and symptoms, laboratory findings, to reach a differential diagnosis for that problem.
- Describe the pathophysiology of the given clinical condition based on the clinical and laboratory findings.
- Discuss the rationale of management, based on the derangement of physiological parameters in a case scenario.

CASE 1.1 IRON DEFICIENCY ANEMIA

A 35-year-old female complains of breathlessness, loss of appetite, apathy and easy fatigability. Examination of the patient revealed pallor, koilonychia, increased heart rate and systolic murmur.

Laboratory investigations of this patient were obtained as under:

- **Hemoglobin:** 6.0 g% (Reference value: 12–14 g% in females; 14–16 g% in males)
- **Packed cell volume (PCV):** 30% (Reference value: 42–45%)
- **RBC count:** 3.0 million/cu.mm (Reference value: 4.5–5.5 million/cu.mm)
- **S. ferritin:** 9 nanogram/mL (Reference value: 12–300 ng/mL)
- **Mean corpuscular volume (MCV):** 70 fL (Reference value: 80–100 fL)
- **Total iron binding capacity (TIBC):** 500 microgram/dL (Reference value: 240–450 microgram/dL)
- **Mean corpuscular hemoglobin concentration (MCHC):** 28% (Reference value: 32–36%)
- **Peripheral blood film (PBF):** RBC are microcytic and hypochromic

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Based on the above case, answer the following questions:

1. What do you think this patient is suffering from?
2. How would you grade the anemia of this patient based on clinical classification?
3. What do you understand about microcytic and hypochromic RBCs in this patient?
4. How is the deficient nutrient absorbed and utilized in our body?
5. Define anemia.
6. Classify anemia according to the etiology (cause).
7. Classify anemia according to the morphology of RBCs.
8. Describe the physiological basis of clinical features/symptoms of this disease.

Provisional diagnosis: Iron deficiency anemia (IDA).
Definition: Decreased oxygen carrying capacity of blood with or without decreased hemoglobin content or RBC count or both.

Classification of Anemia

Etiological Classification

Table 1.1: Etiological classification of anemia.

Main etiology	*Increased blood loss*		*Increased breakdown of RBC*				*Decreased production of RBC*	
Various causes	Acute blood loss	Chronic blood loss	Red cell membrane defects	Enzyme defects	Abnormal hemoglobin	Immune mediated hemolysis	Deficiency of nutrients	Bone marrow disorders
Examples	Acute hemorrhage due to road traffic accident, major surgeries, etc.	Hemorrhoids (piles), hookworm infestation, dysfunctional uterine bleeding in women, etc.	Hereditary spherocytosis, ovalocytosis	Glucose-6-phosphate dehydrogenase (G6PD) deficiency	Sickle cell anemia	Incompatible ABO blood transfusion	Iron deficiency, folic acid/vitamin B_{12} deficiency, protein deficiency	Aplastic anemia, etc.

Morphological Classification

Table 1.2: Morphological classification of anemia.

	Hypochromic (MCHC <31 g/dL)	*Normochromic (MCHC = 31–33 g/dL)*
Size of RBC <7 µm (microcytic; MCV <80 fL)	Iron deficiency anemia thalassemia lead poisoning	Chronic infection
Size of RBC = 7 µm (normocytic, MCV = 80–100 fL)	Chronic hemorrhage	• Acute blood loss hemolytic anemia • Aplastic anemia
Size of RBC >7 µm (macrocytic; MCV >100 fL)	Liver disease	• Vitamin B_{12} deficiency • Folic acid deficiency

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Clinical Classification

Table 1.3: Clinical classification of anemia.

Concentration of Hb	*Grades of anemia*
>10 g/dL	No anemia
8–10 g/dL	Mild anemia
5–8 g/dL	Moderate anemia
<5 g/dL	Severe anemia

Absorption of Iron from GIT

Refer **Figure 1.1**.

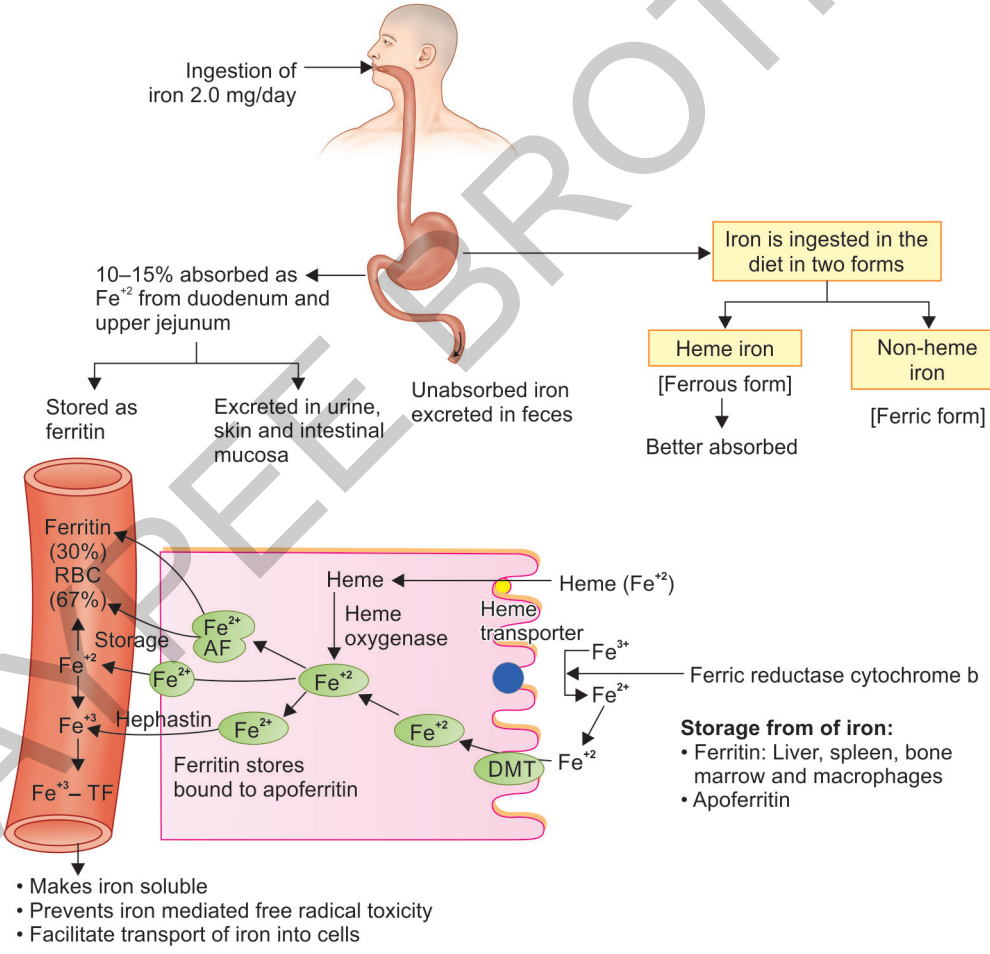

Fig. 1.1: Absorption of iron from GIT.

[DMT: divalent metal transporter; FP: ferro protein transporter; TF: trans ferritin (synthesized in the liver); AF: apoferritin]

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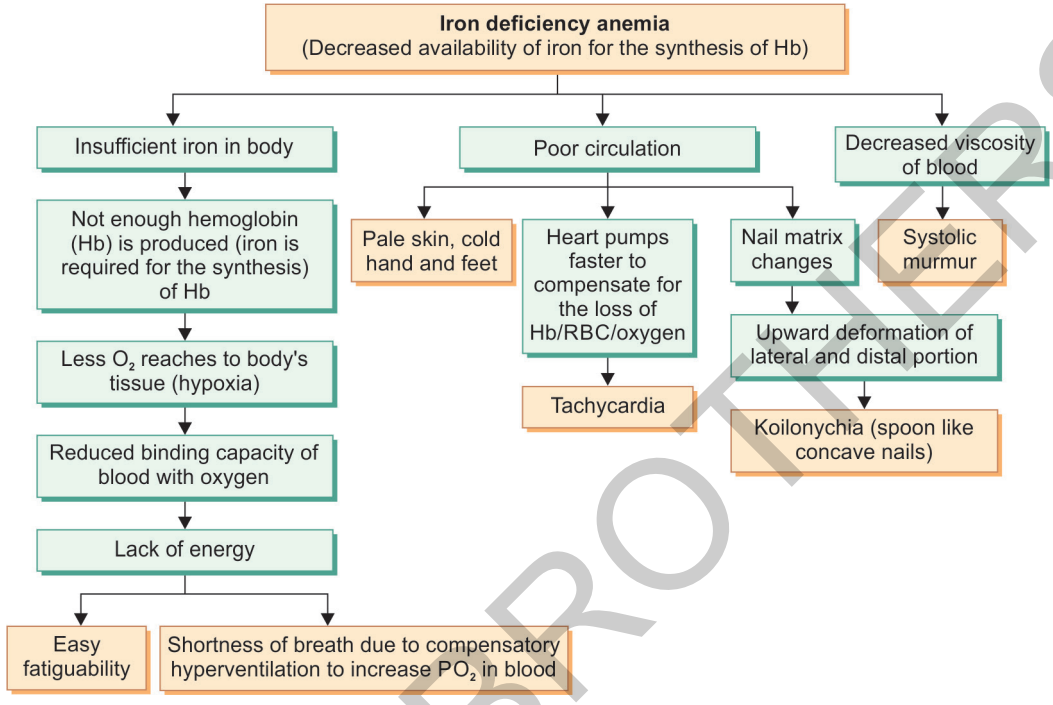

Fig. 1.2: Physiological basis of clinical features of iron deficiency anemia.

Pathophysiology of IDA and Physiology of Clinical Symptoms (Fig. 1.2)

Laboratory Diagnosis

- Decreased RBC count (<4.5 million/cc in females and 5 million/cc in males) and hemoglobin content (<10 g/dL)
- Peripheral blood film shows microcytic hypochromic RBCs
- Red cell indices: MCV <80 fl, MCHC <31 g/dL
- Decreased S. ferritin levels (<10 ng/mL in adult females and <20 ng/mL in adult males)
- Increased total iron binding capacity (TIBC) (>450 µg/mL)

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CASE 1.2 ANEMIA DUE TO VITAMIN B_{12} DEFICIENCY

A 45-year-old male complains of indigestion, diarrhea, soreness in angles of mouth, loss of appetite and tingling sensation in the hands and feet. Examination of the patient revealed angular stomatitis, red beefy tongue and peripheral neuropathy.

Laboratory investigations of these patients were obtained as under:

- **Hemoglobin:** 9.0 g%
- **RBC count:** 1 million/cu.mm.
- **S. Iron:** 170 microgram/dL (Reference value: 60–170 mg/dL)
- **MCV:** 100 fL
- **MCH:** 50 pg
- **Reticulocyte count:** 6% (Reference value: 1–2%)

Based on the above case, answer the following questions:

1. What do you think this patient is suffering from?
2. What is the cause of peripheral neuropathy in this type of anemia?
3. Why do you see large nucleated RBC (megaloblasts) in PBF, in this condition?
4. How is the deficient nutrient absorbed and utilized in our body?
5. Why is this type of anemia seen in patients with gastric atrophy?
6. What are the other conditions with a similar blood picture? What are the causes for the same?
7. What will happen if you give only folic acid supplementation in pernicious anemia?

PBF: RBC are macrocytic and normochromic with some large nucleated RBC, neutropenia with hypersegmented neutrophils and decreased platelets per high power field.

Provisional diagnosis: Megaloblastic anemia (vitamin B_{12} deficiency).

Note: Deficiency of intrinsic factor due to autoimmune causes is termed as pernicious anemia.

Absorption of Vitamin B_{12} from GIT (*Refer* Fig. 1.3)

Both vitamin B_{12} and folic acid, act as methyl carriers and are responsible for the nuclear maturation of the cells, hence present with macrocytic, normochromic and megaloblastic anemia. The cytoplasmic maturation of the cells is normal.

Supplementation of only folic acid, in a patient with vitamin B_{12} deficiency, may result in improvement of blood picture but worsening of peripheral neuropathy. Hence, the patient should be properly evaluated before starting the supplementation.

PBF: The RBCs in peripheral blood film of megaloblastic anemia (both in vitamin B_{12} and folic acid deficiency) are macrocytic and normochromic. The red cells are larger in size but the hemoglobin concentration is normal (increased MCV, normal MCHC). Some of the RBCs are nucleated (seive like nucleus); called as megaloblasts, hence the name ***'megaloblastic anemia'***. The other cells (WBCs and platelets) are also affected showing neutropenia and shift to right on Cooke-Arneth along with thrombocytopenia.

Various causes of vitamin B_{12} deficiency are given in **Table 1.4**.

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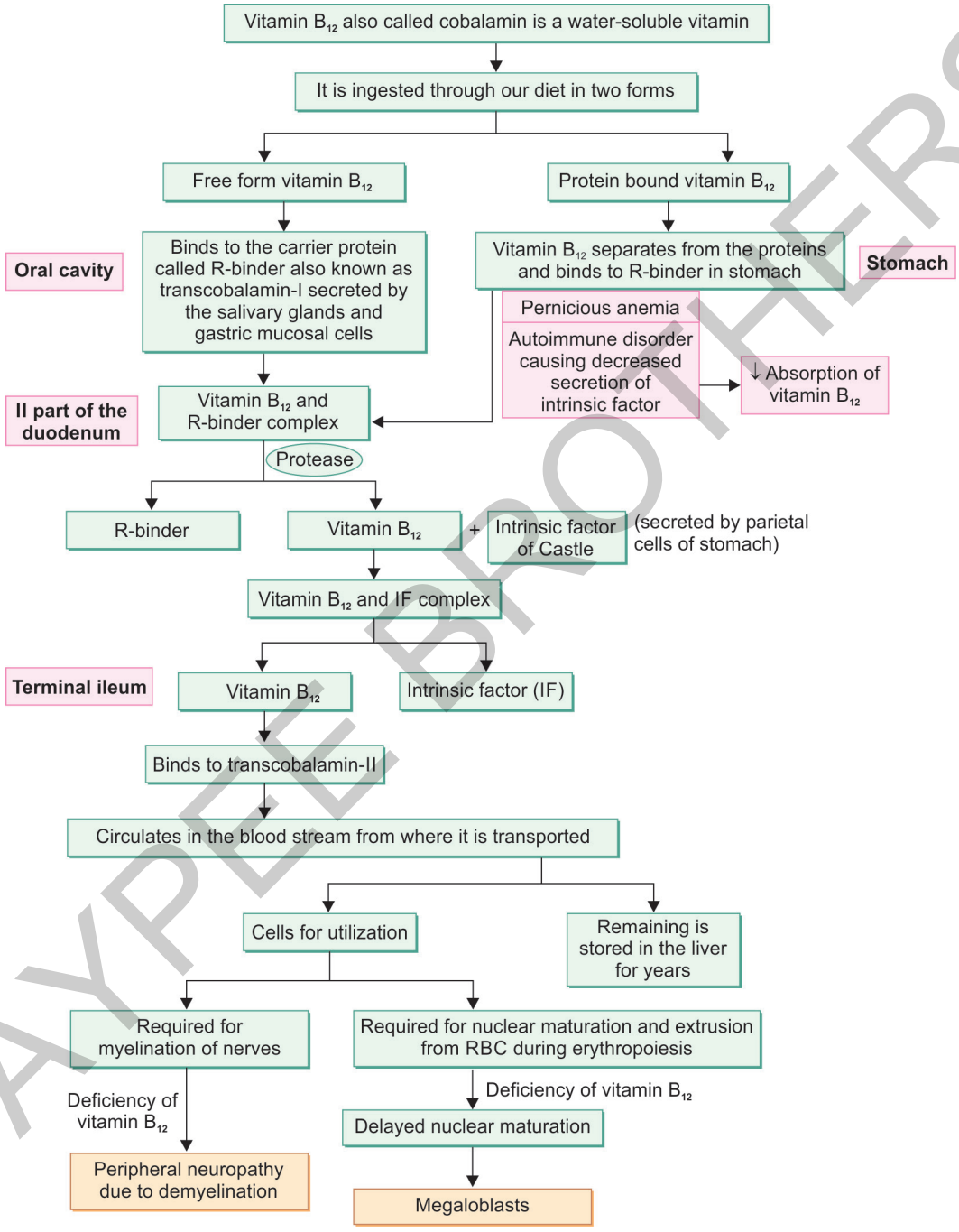

Fig. 1.3: Absorption of vitamin B_{12} from GIT.

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Table 1.4: Various causes of vitamin B_{12} deficiency.

	Gastric	*Intestinal*
Cause	Deficiency of intrinsic factor (IF)	Decrease absorption of vitamin B_{12}
Examples	• Autoimmune causes of failure of secretion of IF (pernicious anemia) • Gastrectomy • Congenital lack of IF	• Tropical sprue • Crohn's disease • Fish tapeworm infestation • Intestinal blind loop syndrome

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CASE 1.3 G-6-PD DEFICIENCY: HEMOLYTIC ANEMIA

A 20-year-old male complains of pain in loin and passage of dark urine after eating fava beans. Examination of the patient revealed nothing significant.

Laboratory investigations of this patient were obtained as under:

- **Hemoglobin:** 10 g%
- **RBC count:** 3 million/cu.mm.
  (***Note:*** Plasma hemoglobin and methemoglobin levels are raised)

PBF: RBC are normocytic and normochromic with polychromasia, basophilic stipplings and marked poikilocytosis.

Based on the above case, answer the following questions:

1. What do you think this patient is suffering from?
2. What are the causes of this type of anemia?
3. What is the mechanism of action of the deficient enzyme in this condition?
4. How can you prevent this condition?
5. What are the other types of hemolytic anemia and what is the cause in each one of them?
6. What are target cells? In which condition, they are seen?

Provisional diagnosis: Hemolytic anemia (G-6-PD deficiency).
Hemolytic anemia could occur due to many causes, as given in **Table 1.5**.

Pathophysiology of G-6-PD Deficiency and Physiology of Clinical Symptoms

See **Figure 1.4**.

Table 1.5: Causes of hemolytic anemia.

Due to defects within the RBCs (intracorpuscular defects)	*Due to various pathologies outside the RBCs (extracorpuscular defects)*
Defects in red cell membrane: • Hereditary spherocytosis • Hereditary ovalocytosis	Mechanical trauma to RBCs: • Thrombi within circulation • Exotoxins
Defective hemoglobin chains (hemoglobinopathies): • Thalassemia • Sickle cell anemia	Antigen antibody mediated hemolysis: • ABO incompatibility • Rh incompatibility • Drug induced
Enzyme defects: • G-6-PD deficiency • Pyruvate kinase and hexokinase deficiency	

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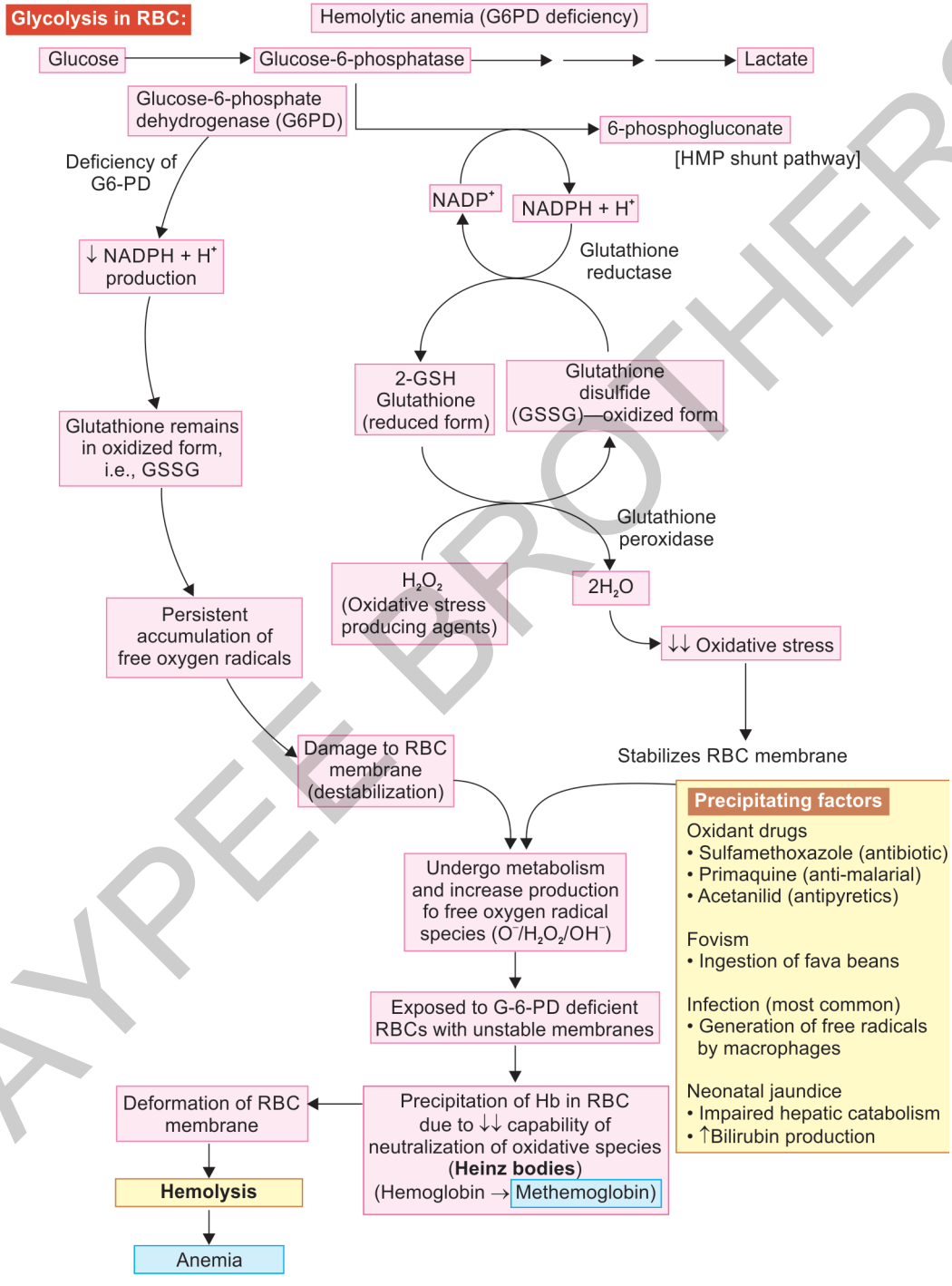

Fig. 1.4: Pathophysiology of G-6-PD deficiency anemia.

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CASE 1.4 ERYTHROBLASTOSIS FETALIS

A 4-day-old home delivered male baby was brought to the pediatrician with complaints of excessive cry and jaundice. The mother of the baby is from a village and did not avail any antenatal check-up during the pregnancy. She has a 2-year-old male child who was a full term normal home delivered baby.

Mother has no history of abortion and no history of jaundice in the first baby after birth. Details of the blood group of the family are not available.

On examination: Baby is lethargic with poor muscle tone, poor feeding and jaundice.

Investigations:

- **Mother:** Blood group is B negative.
- **Baby:**
  - *Blood group:* AB positive
  - *Hb:* 10 g%
  - Erythroblasts: Abundant (erythroblastemia)
  - *S. bilirubin:* 18 mg/dL (Reference value: 0.2–0.8 mg/dL)

Based on the above case, answer the following questions:

1. What is the probable diagnosis?
2. What is the pathophysiology behind the clinical condition presented?
3. How can this condition be prevented in subsequent pregnancies?
4. What is the physiological basis of prophylaxis given?

Provisional diagnosis: Hemolytic disease of newborn due to Rh incompatibility (erythroblastosis fetalis).

Pathophysiology of Hemolytic Disease of Newborn (HDN) (Fig. 1.5)

The first pregnancy can also be affected by Rh incompatibility, in case of previous exposure to D-antigen (as in case of transfusion of Rh positive blood to Rh negative lady).

The concentration of anti-D antibodies formed in the mother will depend upon the antigen load entering the maternal circulation.

Based on the severity of HDN, it is classified as:

1. **Hydrops fetalis:** Most severe. Baby is swollen and baby dies in utero or few hours after birth.
2. **Icterus gravis neonatorum:** Characterized by hemolytic jaundice, anemia, erythroblastemia, kernicterus (deposition of bilirubin in basal ganglia, if more than 18 g%) and liver damage.

Prophylaxis: Injection anti-D (RhoGam) within 72 hours of delivery. It is a type of passive immunity.

Treatment:

1. Intrauterine fetal blood transfusion
2. Exchange transfusion: Transfusing Rh negative blood of same ABO type to stop hemolysis, till the antibodies persist in neonatal blood.
3. Phototherapy
4. Immunoglobulin therapy

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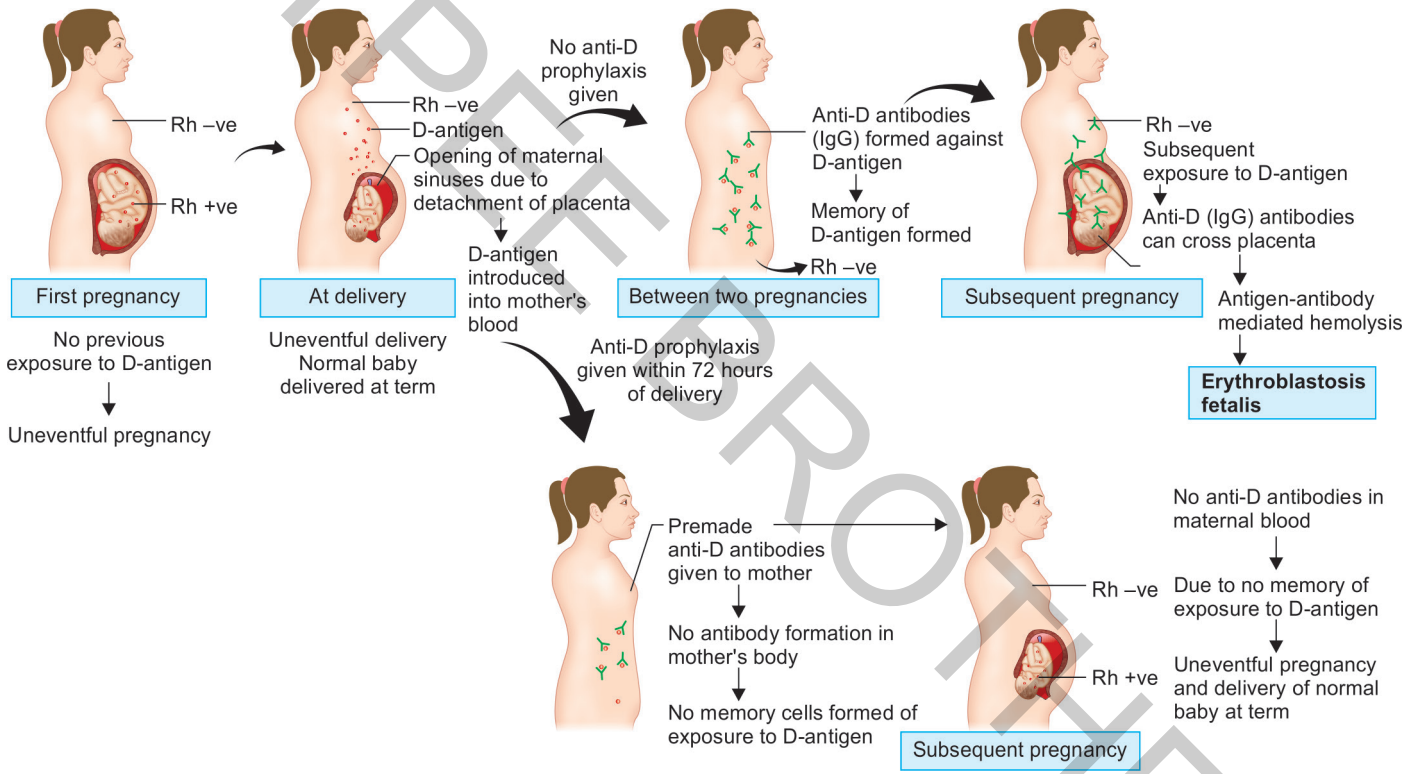

Fig. 1.5: Pathophysiology of hemolytic disease of newborn.

Note: Erythroblastosis means the increased number of circulating erythroblasts.

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CASE 1.5 HEMOPHILIA

A young boy of 8-year-old complained of a swollen knee after he fell from the bicycle. He also complained of some bluish discoloration of the skin of right arm. His past history is suggestive of prolonged bleeding even after minor injury. On examination, he was found to have a hematoma of the knee. His laboratory investigations show prolonged CT, APTT with normal BT and PT. His hemoglobin is 9.0 g%.

Based on the above case, answer the following questions:

1. What is the probable diagnosis?
2. Why is the bleeding time normal in this patient despite a prolonged CT and APTT?
3. What is the physiological basis of the clinical symptoms?

Provisional diagnosis: Hemophilia A

Hemophilia is a bleeding disorder and is of two types:

1. **Hemophilia A:** Due to deficiency of factor VIII (more common)
2. **Hemophilia B:** Due to deficiency of factor IX

Hemophilia A is a clinical condition that arises due to deficiency of factor VIII resulting in bleeding tendencies.

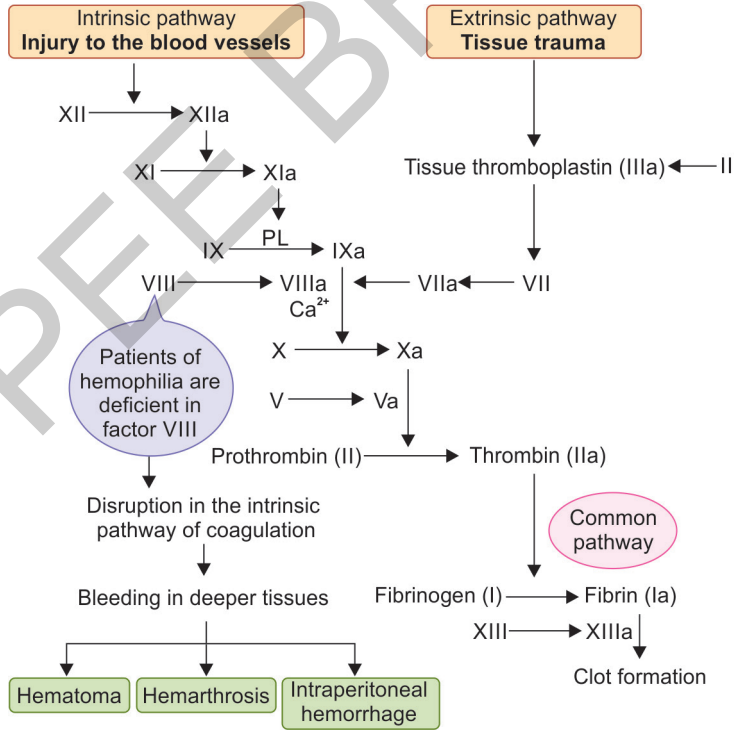

Fig. 1.6: Pathophysiology of hemophilia A.
(PL: phospholipid)

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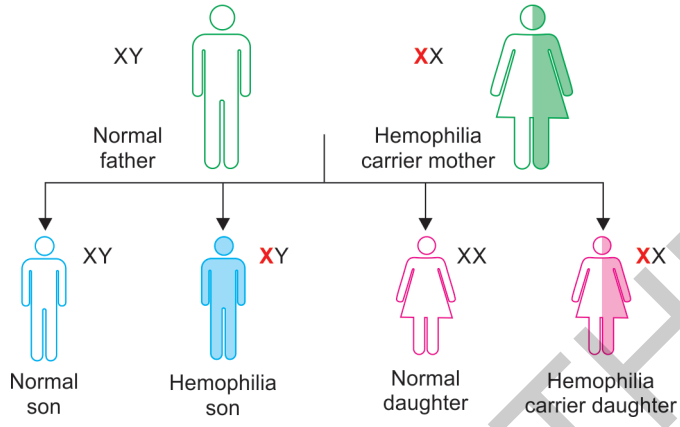

Fig. 1.7: Inheritance in hemophilia A patients.

Factor VIII is present on the X- chromosome and hence it is inherited as a X-linked recessive genetic disorder.

Diagnosis of hemophilia begins with a good clinical history and physical examination supported by the following laboratory investigations:

1. **Bleeding time (BT):** Normal (as platelet count is normal, temporary hemostasis is not affected).
2. **Clotting time (CT):** Prolonged (due to derangement in the intrinsic pathway of coagulation).
3. **Prothrombin time (PT):** Normal
   - PT is a test used for extrinsic pathway and common pathway of coagulation (factors involved are VII, X, V, II, I)
4. **Activated partial thromboplastin time (APTT):** Prolonged.
   - Test is used for intrinsic and common pathway of coagulation (factors involved are XII, XI, IX, VIII, X, V, II, I)
   - Factor VIII and IX are a part of intrinsic pathway, so APTT is increased in hemophilia A and hemophilia B).

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CASE 1.6 SICKLE CELL ANEMIA

A three-and-a-half-year-old male child was brought to the OPD with complaints of refusal to eat and off and on pain in the abdomen. He also complained of off and on joint pains.

On examination: The child was cooperative but irritable, well oriented to time, place and person.
Vitals: Normal.
Pallor and icterus: Present
Abdominal examination: Splenomegaly
CVS and respiratory system: NAD
Laboratory investigations:

- **Hb:** 8 g%
- **RBC:** 3 million/cmm
- **PCV:** 30%
- **MCV:** 86 fL
- **MCHC:** 32%
- **Peripheral blood film:** Normocytic normochromic RBCs
- **S. bilirubin:** 2.8 mg/dL
- **Hb electrophoresis:** SS pattern
- **Solubility test also known as sickling test:** Showed sickle cells
- **High performance liquid chromatography (HPLC): technique:** Presence of HbS

Based on the above case, answer the following questions:

1. What is the probable diagnosis?
2. What is the pathophysiology of the given disease?
3. What is sickle cell crisis?
4. How does it differ from thalassemia?

Provisional diagnosis: Sickle cell anemia

Definition: It is a genetic disorder characterized by the presence of HbS instead of Hb A.
Pathophysiology:

	Normal person	*Person with sickle cell anemia*
DNA	CTC	CAC
mRNA	GAG	GUG

It is a type of point mutation in the gene that encodes for the beta-globin chain of hemoglobin. It leads to formation of an abnormal Hb, known as HbS. In HbS, at the 6th position of the β-chain, glutamate (which is normally present in HbA) is replaced by valine.

Clinical presentation

- Occlusion of the microvasculature

  ↓

  Ischemic pain and necrosis of tissue distal to the occlusion
- Features of hemolytic anemia
- Features of hemolytic jaundice

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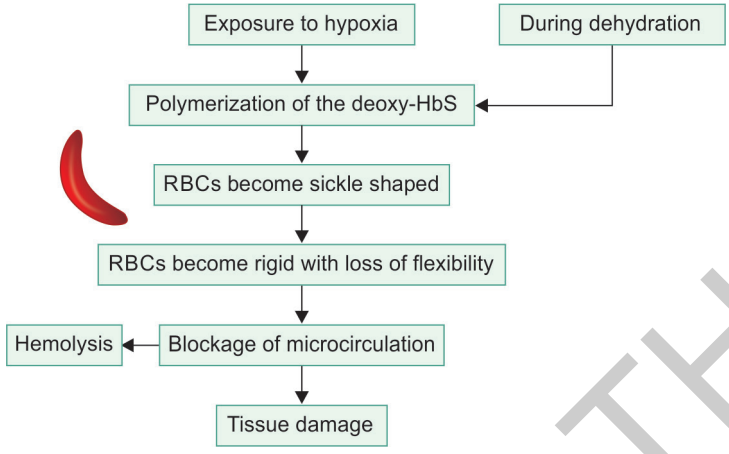

Fig. 1.8: Pathophysiology of sickle cell anemia.

- Hepatosplenomegaly
- **Vasoactive crisis:**
  - Stroke
  - Ischemic necrosis
  - Acute chest syndrome

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