Pattern 3: Hereditary Spherocytosis

HS may present during anytime from the neonatal period to adulthood. When there is a family history, it is usually easy to suspect the diagnosis.

- History strongly indicative of chronic extravascular hemolysis, usually but not always **hereditary spherocytosis**: normal patient presenting with **pallor, mild jaundice, an enlarged spleen (classic triad)** and a family history of a relative who had been "cured" of the same problem by splenectomy. Also family history of **pigment gallstones** in a young person.
- Peripheral smear with **spherocytes and raised osmotic fragility** test are features of HS.

Case No. 7

History: A 24-year-women complains of chronic fatigue since childhood and mild icterus. On examination she is **anemic, jaundiced and has moderate splenomegaly**. Her mother had similar complaints and has multiple **pigment gallstones**.

Inference (cases No. 7 and 8): mild jaundice, anemia and splenomegaly constitute a classical triad, and along with family history/pigment gallstones favor hereditary spherocytosis.

Case No. 8

History: A 15-year-old girl presents with weakness and fatigue. On examination, her sclera shows mild jaundice and mild splenomegaly. **Family history** indicated about her father having recurrent gallstones.

Positive osmotic test fragility confirms the diagnosis of HS.

Laboratory findings for cases 7 and 8: Hb 11.1 g/dL, RBC count 3.6 million/cumm. WBC count 6,800/cumm and platelet count 1,75,000/cumm. MCV is 78 fL, MCH is 32 pg and MCHC is 38 g/dL. In a laboratory test, her **RBCs lyse at a higher concentration of saline (osmotic fragility test)** compared to normal patients.

Pattern 4: Thalassemia Major

- History strongly indicative of **thalassemia major: short stature, failure to thrive, severe pallor, jaundice,** maxillary over-growth (bossing), **frontal bossing**, sallow complexion and splenomegaly.
- Laboratory finding of **microcytic hypochromic anemia** with **many target cells, reduced MCV,MCH and MCHC, increased serum iron normal RDW** are features of thalassemia major.

Case No. 9

Inference (case No. 9): failure to thrive, severe pallor, jaundice, splenomegaly, microcytic hypochromic anemia with target cells, reduced MCV and normal RDW points to the diagnosis of β-thalassemia major. Increased iron stores and raised serum ferritin is against the diagnosis of IDA (refer Table 3.3).

History: An 11-month-old male child was brought to the pediatric outpatient by his parents and complained that the child was **failing to thrive**. On examination, jaundice, **pallor** and a **palpable spleen** was detected.

Laboratory findings: Hb 7.8 g/dL, hematocrit 23.4%, **MCV 66 fL,** platelet count 1,75,000/cumm, and WBC count 8,200/cumm. His **serum ferritin was 3250 ng/mL**. Peripheral examination showed **microcytic hypochromic anemia** with many **target cells.** A bone marrow aspiration performed and reveals a myeloid: erythroid ratio of 1:4, and increased iron stores.

Pattern 5: Sickle Cell Anemia

- History strongly indicative of **sickle cell anemia: recurrent episodes of vaso-occlusion** in connective and musculoskeletal structures anywhere in the body producing ischemia and manifesting as **acute pain**, **tenderness and fever**. Child may show generalized impairment of growth and development. **Adults** manifest with **chronic organ damage**.

Inference (case No. 10): the recurrent episodes of pain abdomen and backache points to a vaso-occlusive crisis in sickle cell anemia.

- Presence of **sickle cells** in the peripheral smear, **positive sickling test**, along with demonstration of HbS by **hemoglobin electrophoresis** and **HPLC** is diagnostic of **sickle cell anemia**.

Case No. 10

History: A 10-year-old boy complains of **severe pain** in the **chest, abdomen and bones**. On enquiry, his mother reveals that he had **several episodes** of severe abdominal and back pain since early childhood. Physical examination shows **anemia, jaundice and leg ulcer.**

Laboratory findings: Hb 11.0 g/dL, RBC count 3.2 million/cumm, WBC count 8,800/cumm and platelet count 1,95,000/cumm. Peripheral smear examination, **hemoglobin electrophoresis and HPLC** confirms the diagnosis.

Presence of sickle cells in the peripheral smear and presence of more than 70% Hb S on hemoglobin electrophoresis is confirmative.

Pattern 6: Immune Thrombocytopenic Purpura (ITP)

- History strongly indicative of **acute immune thrombocytopenic purpura**: sudden development of bruising, petechiae and **muco-cutaneous bleeding** in a **child following a bacterial or nonspecific viral** (upper respiratory or gastrointestinal) **infection** without features of acute leukemia (such as anemia and bone tenderness).
- Laboratory findings of **markedly reduced platelet count** and **megakaryocytic hyperplasia with immature megakaryocytes.** In the bone marrow favors the diagnosis of **acute immune thrombocytopenic purpura**.

The severity/nature of bleeding depends on the platelet count.

Case No. 11

History: A **14-year**-male comes to the OPD with **petechial rashes of 2 days** duration. He has no history of bleeding in past. He had an attack of **viral infection 3 weeks before** this complaint.

Laboratory findings: Hb 14.5 g/dL, hematocrit 45%, RBC count 5.1 million/cumm. WBC count 7,200/cumm and **platelet count 75,000/cumm**. MCV is 85 fL, MCH is 32 pg and MCHC is 31 g/dL and RDW is 14.

Inference (case No. 11): petechial rashes of short duration in a child, low platelet count is due to acute idiopathic thrombocytopenic purpura.

Pattern 7: Hemophilia

- History strongly indicative of one of the **hemophilias (A or B)**: a **male child** presenting with a serious bleeding disorder, particularly with **hemarthrosis**.
- **Decreased factor VIII/IX and increased APTT is diagnostic of** one of the **hemophilias (A or B).**

Case No. 12

History: An **18-year-male** complains of **knee joint swelling** after minor trauma. On examination, the joint appears tense, red and swollen. **Family history** revealed similar complaints by his uncle.

Case No. 13

History: A **6-year-old boy** gives a history of **easy bruising** and episodes of passing blood in urine since infancy. On examination, many ecchymoses are noted in the skin of lower limbs. **Family history** reveals similar complaints in members of the family involving only males and history of **hemarthrosis** in few of them.

Inference (cases No. 12 and 13): the development of ecchymosis, family history with disease affecting males, hemarthrosis, normal platelet count and prolonged APTT favor hemophilia A/B.