

Case 1:

- A 24 years old female complains of dizziness, fatigue, shortness of breath especially on exertion (not while resting) and Headaches for the last 2 months (She has anemia syndrome). She has been losing scalp hair (common complaint in iron deficiency anemia). She does not eat red meat (iron is an essential component of blood cells; red meat is a great source for iron) and has reported heavy menstrual bleeding (causes of anemia are both dietary and blood loss). Her physical exam showed pallor in the skin (to test for pallor we look at the skin on the palms and the conjunctiva; they are at the body's core temperature (36.8)). Nails are bent and pointing downwards. We can see chylosis and angular stomatitis. All these symptoms point toward iron deficiency anemia. She has dysphagia, as well (food gets stuck while swallowing, painful swallowing). Upper endoscopy revealed an esophageal web (a folding in the mucosa of the esophagus; it produces symptoms related to swallowing and is commonly found in iron deficiency anemia patients. If the patient takes a "barium swallow", you will find that this area does not receive contrast (looks like a black line in 2D)
- The most likely diagnosis here is Iron deficiency anemia.
- Lab tests revealed:
 - o Moderate anemia
 - o very low MCV
 - o high RBC
 - o MCH is relatively low...
 - o Blood film is hypochromic
 - o Anisocytosis: Cells don't look the same. It is a very important finding in Iron deficiency anemia.
 - o Poikilocytosis: odd looking cells (rod-like or pencil like)
 - o Ferritin is low. Ferritin measurements reflect the amount of stored iron in the body. A single test is enough to indicate if this patient is iron deficient.
 - o Low saturation of iron binding protein
 - o Serum Fe is low.
 - o B12 and Folate are normal indicating that this is an isolated iron deficiency anemia secondary to poor diet and bleeding.
- Now that you know the disease, you have to investigate the causes of menorrhagia.
- RDW: In a normal patient, most cells would be clustered around a narrow range. In patients with iron deficiency anemia, and due to anisocytosis there is a high RDW. It is a strong indication for iron deficiency anemia. If you take a blood film that is hypochromic, microcytic with high RDW, the first thing you should think about is iron deficiency anemia. For hair to grow and for it to be maintained healthy, it needs iron.

Case 2:

- 65 years old male had gradual onset of “odd” behavior (layman terms) with psychotic symptoms. He was noticed to have imbalanced gait. (Since the dominant symptoms are neurological, we have to test for neural symptoms). We use the vibration test using a tuning fork. proprioception in lower limbs is lost; this is due to loss of sensation in thick nerve fibers responsible for this sensation. These get affected due to B12 deficiency (many other conditions might cause this symptoms, but this is the most important) . Lab tests revealed Hb 5 and MCV 112. As you notice, this patient does not have anemia symptoms; this is due to the gradual onset.
- Lab tests:
 - o MCV is very high
 - o WBC and platelet count on the low side
 - o Reticulocytes count corrected is an important measure. It needs to be corrected depending on the case of anemia.
 - o Serum B12 is low
 - o LDH is very high. If there is any destruction in any cell in the body, LDH will increase dramatically; it is nonspecific.
- This is probably B12 deficiency anemia. In order to prove it, you have to check for HCl deficiency in the stomach; HCl is important for B12 absorption. Without it, you don't absorb B12.
- Physical examination reveals skin depigmentation (vitiligo), an enlarged tongue due to loss of mucosa, stomach biopsy shows abnormal mucosa (absence of oxyntic gastric cells which produce HCl). Additional serum tests show intrinsic factor and parietal cells antibodies. This condition is an autoimmune condition. This is a disorder where you produce antibodies against gastric mucosa causing its atrophy. Some of these antibodies might be directed towards the skin causing depigmentation.
- The blood film shows ovalocytes that are not hypochromic, but they are large. We have hyper-segmented neutrophils, and red cells are somehow elongated. We also have chromocytopenia. The blood film also reveals nucleated cells. These cells are typically found in the bone marrow, but, here, we see them in the blood.
- **This is pernicious anemia: B12 deficiency secondary gastric cause autoimmune condition**

Case 3

- 18 years old male complains of acute pain in his back, Dizziness, Fatigue, Shortness of breath and Headaches for the last 6 hours (anemia syndrome with pain in the back). This is a chronic condition. We have pallor, jaundice, ulcer in the internal urogenitals, and Abnormal fingers.
- Blood film shows hypochromia and odd looking cells (sickle cells)

- X-ray of the spine shows that the vertebrae are crushed (mouthfish vertebrae; characteristic of sickle cell disease; they are caused by vertebrae fractures in the spine)
- Lab tests:
 - o Hb 9 so he is anemic
 - o MCV is a bit low... hypochromic
 - o RDW is about normal
 - o Sickle cell test was positive (A 0%, A2 3%, S 90%, F 7%)
 - o He has a homozygous sickle cell disease.
 - S-S → disease
 - S → sickle cell trait
- **His diagnosis is sickle cell disease with vasocclusive crisis.** When Hemoglobin is deoxygenated, it becomes insoluble making a polymeric insoluble substance which bends the red cells. These cells block the small venules and arterioles causing infarcts in that area.

Case 4:

- 13 years old male complains of skin pigmentation, abdominal swelling and pallor. He has been receiving blood transfusions since the age of 9 months. Physical examination reveals hyperpigmented skin, protruding upper jaw, protruding frontal bone and cheeks, very large spleen, and stunted growth
- Skull X-ray and CT scan of the skull: normally, the bone looks like a half circle; but here you notice that the bone is deformed like he is wearing a helmet. It looks like there is hair on the rear end of the skull. In adults, the skull is an important source for hematopoiesis. All flat bones are places for hematopoiesis. This enlargement reveals an enlargement of the bone marrow compartment of the skull
- Blood film: hypochromia, microcytosis, target cell, anisocytosis, nucleated red blood cells (abnormal; it indicates replacement of bone marrow or hydrosis of bone marrow), very high ferritin (6000), most Hb is HBF (A 25%, A2 6%, F 69%)
- Diagnosis: **B-Thalassemia major with Fe overload.** Iron overload is what usually kills thalassemia patients! It causes multisystem failure; especially heart and liver failure

Case 5:

- 16 years old male complains of back pain, dizziness, red urine and pallor after eating fava beans (favism) He had similar attacks before.
- Physical exam shows pallor and tachycardia.
- Lab tests indicate an acute attack (change of color in the test tubes). Urine was taken and centrifuged, then stained to test for iron. You see hemosidren granules, which are iron granules found in renal tube cells.

- Blood tests show: Low hemoglobin, low haptoglobin (hemoglobin when in circulation first binds to haptoglobin, then albumin then becomes free circulating haptoglobin.), Serum LDH is 1400, high Bilirubin is high (mostly indirect)
- G6PD cannot be measured on the same admission; you have to wait for two months.
- Tests show low G6PD and Heinz bodies (Heinz bodies indicate denaturation and precipitation of Hb. When the red blood cell passes through the sinusoids it is chopped off.)
- Diagnosis: **severe type of favism**

Case 6

- 50 years old man complains for several weeks of hotness in his face, itching and severe acute pain in his big toe.
- Physical examination reveals distended blood vessels. The right big toe is swollen indicating an acute attack of gout.
- Investigations: Hb 19 (high), WBC 17000(high), Retic 500K (high), Uric acid is 12 (high; you can accept up to 7), PO₂ saturation is normal, serum erythropoietin is normal
- Molecular biology tests reveal a JAK II mutation (very common in myeloproliferative diseases)
- Diagnosis: **polycythemia vera with acute gout**

Case 7

- 19 years old boy complains of repeated attacks of large joint painful swelling especially in his knees for several years. His maternal uncle has similar condition (familial condition)
- Physical examination: the knee is swollen with deformity and wasting of the muscles and soft tissue bleeding in the abdomen.
- Lab tests show that PT is normal, PTT is prolonged indicating something wrong in the intrinsic pathway or common pathway of coagulation, TT is normal, BT 7 (used to indicate platelet function), factor 8 less than 1%, Factor 9 100% (normal)
- Diagnosis: **severe hemophilia A**. Hemophilia A and B are clinically indistinguishable; laboratory diagnosis is the indication

Case 8

- 49 years old lady complains of painful swelling and hotness of her L leg following coming back from visiting her relatives in USA. She had repeated attacks of cough with hemoptesis and shortness of breath.
physical examination revealed a tender swollen leg and thigh indicating that the block is higher up
- CT scan shows obstruction indicating a pulmonary thrombus blocking blood vessels. This was confirmed by a duplex ultrasound.

- Many of those who develop DVT, have a deficiency in one of the antithrombotic factors. Protein C is the most important; Protein C deficiency is a physiological anticoagulant. It is related to factor 8 and 5

Case 9

- 29 years old lady complains of fever and painful gums for 1 week. She developed easy bruising and hemorrhagic spots on her trunk.
- The most important signs are infection and bleeding; the lungs severely hypertrophic with bleeding, hemorrhagic spots on the thigh
- Lab tests: Hb is low, high WBC high platelets, low PT, low PTT, LDH is high, and bone marrow infiltrated by blasts
- Diagnosis: **AML**

Case 10

- 69 years old man complains of fever and cervical and axillary swelling for several months with occasional fever and productive purulent cough(indicates some sort of bronchitis)
- Physical examination reveals enlarged axillary lymph nodes, large tonsils, and enlarged cervical lymphs
- Lab tests show Hb 10 mild anemia, WBC count is very high, platelets are high
- Blood film shows mature lymphocytes (most of the cells are lymphocytes)
- Diagnosis: Chronic lymphocytic leukemia

Case 11

- 57 yr old man complains of back pain for several months and fracture of his L leg 2 days ago. An X-ray of his thigh was taken to confirm the fracture. Skull X-ray shows black spots (osteolytic lesions). We can see an infiltration of skin and subcutaneous tissue in the forehead area. Bone marrow shows plasma cells with a peripheral nucleus blue colored and binucleated. His lab tests reveal that he is anemic
- Blood film shows rouleaux cells (put on top of each other)
- Electrophoresis shows an abnormal substantial increase in the IgG kappa region
- Giving us the diagnosis of - Multiple myeloma IgG/κ stage III

Questions:

- 1) A female complaining shortness of breath at exertion... anemia syndrome. She may have all the following sign except?
 - a. Iron deficiency
 - b. Scalp hair loss
 - c. **Loss of vibration sense**
 - d. Angular stomatitis
- 2) Which of the following is characteristically part of this clinical image? (hemophilia)
 - a. **Large soft tissue bleeding following minimal trauma**
 - b. Recurrent mucosal bleeding
 - c. Prolonged bleeding time