

Aplastic Anemia Cases. MZ.

Case 1



- 6yo male
- History of Easy bruising
- Presents with fatigue, CBC - aplastic anemia

- On PE:
 - 25% for height
 - Several bruises
 - Café au lait spots
 - Weak radial pulse



Workup

- Bone marrow – hypocellular
- Chromosome of phytohemagglutinin-stimulated lymphocytes: increased numbers of spontaneous breaks.
- Over the next year, the blood counts gradually decreased, and cell counts responded to oxymetholone.
- What's your Dx?

Fanconi's Anemia

- Auto Recessive; 10-20% of families with consanguineous marriages
- Pancytopenia (4-12yrs). Macrocytosis, high HbF, high erythropoietin are characteristic of stress erythropoiesis.
- Characterized by diepoxybutane-induced chromosomal breakages
- BM: hypocellular
- PE: café au lait spots, short stature – impaired GH secretion, radial and long bone abn, hypogenitalism, microcephaly, eye probs(stabismus, microphthalmia)
- Higher risk for Cancers

Case 2



- 14yo girl presented to the ER with heavy menstrual periods, and severe epistaxis.
- PE notable for petechiae and bruising
- CBC – pancytopenia with 0.1% retics
- BM aspirate was very hypocellular showing mainly fat.

Case 2

- Her menorrhagia and epistaxis ceased with PRBC transfusions, platelets and oxymetholone
- Following hospital discharge, her blood counts did not improve. She developed a high fever and died from Gm(-) sepsis.
- What's your Dx?

Acquired Aplastic Anemia

- Affecting 1-5mill children per year. No sex predisposition, no age peak.
- In 1/2 the cases no cause is found
- In past, chloramphenicol is most implicated.
 - Other agents: sniffing glue, antibacterials (sulfa), phenytoin, chlorpromazine, thiouracil, methicillin. Insecticides, Toxins (benzene)
 - ID (Hepatitis, HIV, mono, influenza, MMR, RMSF, CMV, HSV)

Case 3

- Hyperpigmentation of the face, neck, shoulders
 - Dystrophic nails,
 - Mucous membrane leukoplakia
 - Aplastic Anemia
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- What's the Dx?

Dyskeratosis Congenita

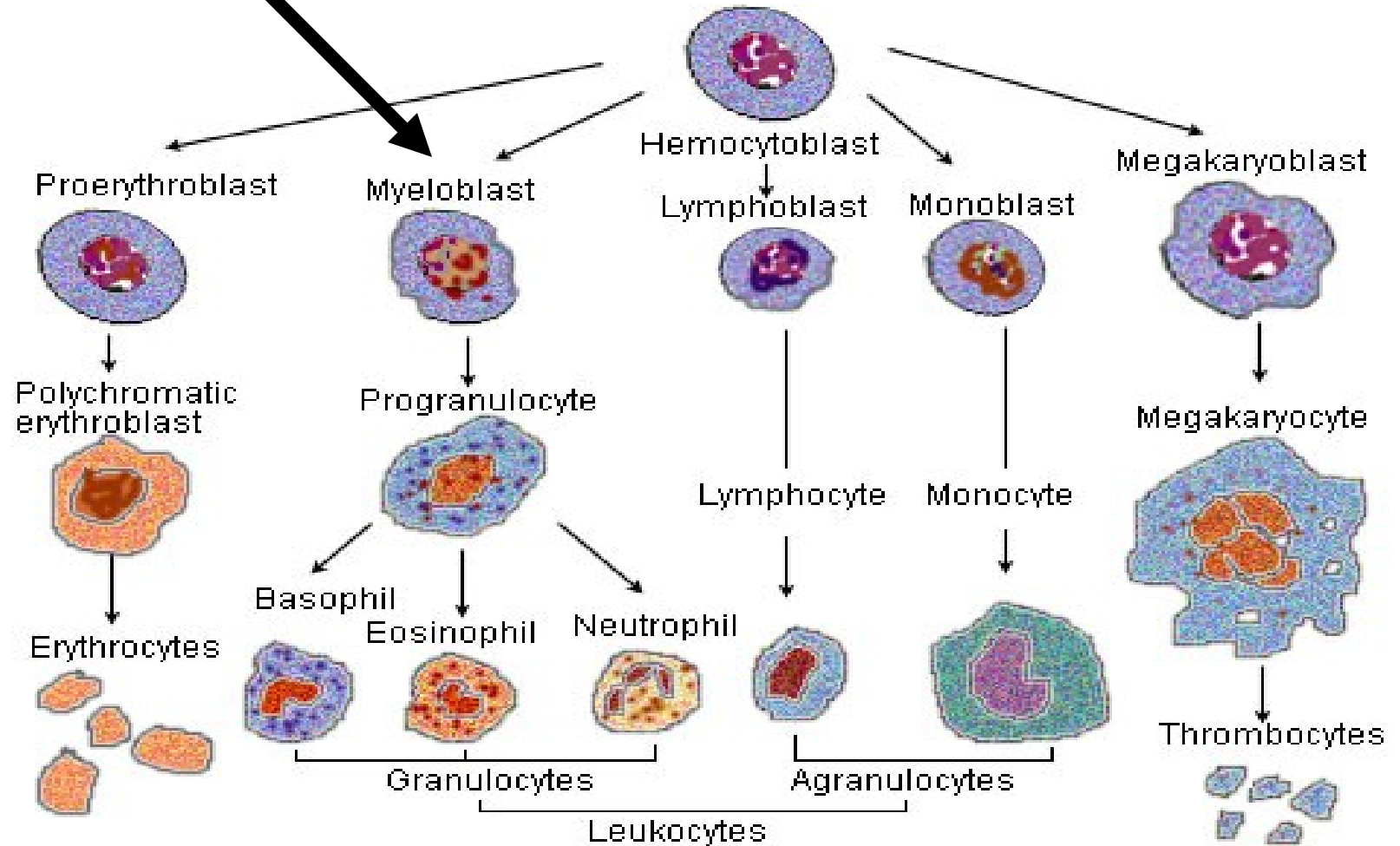
- X-linked type of extodermal dysplasia
- Bone marrow failure usually begins with macrocytic anemia
- Transiently responsive to androgen therapy.

Case 4



- 3yo boy develops pallor and bruising, and 3wks of intermittent fever.
- Pancytopenia found on work up without blasts in peripheral blood

Case 4



What's the Dx?

AML

- This case emphasizes the blasts are not always seen on the smear – especially when leukemia presents with low WBC counts.

Case 5

- 13yo girl with 1wk of low grade fevers and fatigue, and sore throat
 - PE: nontender anterior cervical lymph nodes
 - Enlarged spleen 3cm below the costal margin.



Case 5

- EBV titers are positive, and she was given a dose of steroids.
- Two days later, she presents to the ER with no urine output for 18hrs.
 - CBC in the ER reveals WBC count of 135000 with 95% blasts

What do you think happened?

Case 5

- Steroid induced tumor lysis in a patient who has ALL
- Hemodialysis was initiated to correct the electrolyte abnormalities.
- Oral urate oxidase was administered to block uric acid formation
- Aluminum hydroxide was administered orally to bind phosphate.