04/07/21 Morning Report with @CPSolvers

Case Presenter: *Han Nguyen(@Flower_Freeland)* ***Case Discussants***: *Amanda Barreto (@amandabarretof2) and Kaitlyn Thomas (@smalltownOMS)*

CC: epistaxis

HPI: 13F presents to ED w/ spontaneous, unremitting epistaxis
No recent injury or trauma.
ROS: No fever, fatigue, headache, vomiting, nausea, abdominal pain, hematuria

PMH:
7 yo: skin pigmentation & nail dystrophy
Thrombocytopenia on unknown tx
Nl menses

Meds:
None

Fam Hx:
None

Soc Hx: Student, lives in Vietnam, no recent travel

Health-Related Behaviors:
No tobacco, alcohol, other drugs

Allergies: NKA

Vitals: **T**: **HR**: **BP**: **RR**: **SpO_2**: nl **Ht:** 152 cm, **Wt:** 40 kg
Exam:
Gen: awake, well appearing
HEENT: pale conjunctiva, smooth tongue w/loss of papillae
CV, **Pulm**, **Abd, Neuro:** nl
Extremities/Skin: Dystrophic nails in both hands & feet, hyperpigmented macules, ecchymosis in arms & legs

Notable Labs & Imaging:
Hematology:
WBC: 2.1 (ANC 580) Hgb: 9.1 MCV: 90.4 MCH 28.3 pg RI: 1.68 Plt: 7000 Imm frax 12.2%
Chemistry:
Na, K, Cl: nl
BUN: 8.57 Cr: 0.69 glucose: 95
AST: 25 ALT: 51 GGT 35 T. Bili: 0.94 D Bili 0.18 LDH 265 Haptoglobin 0.12
Iron: 125.6, Transferrin: 303.3, TSat: 29.5%, Ferritin: 45.86 B9: 3.3, B12: 249
Coags: PT 13.6, PTT 33.4, Fibrinogen 144.9 TSH: 0.92, fT3: 3.09, fT4: 0.95
Smear: hypochromic, anisocytosis, thrombocytopenia
ANA, anti-dsDNA, LE cell, AntiSm, APLS, Direct/Indirect Coombs: (-)
HBV, HCV, HIV: (-) EBV IgM(-)/IgG (+), CMV IgM (-)/IgG (+)
CD55/59: nl
Hb electrophoresis: A: 94.4%, A2:2.5%, F: 3.1%
BMBx:
Aspirate: Hypercellular, erythopoeisis increased, metamyelocytes incr.
1st: Cellularity: 70%, M/E ratio=0.5, Trilineage hematopoesis w/nl maturation & no dysplasia
Others: abnormalities in nuclei of some erythrocytes, presence of histiocytes
PERLS stain: Grade 0, Sideroblast <5%
2nd: 8 samples w/2% cellularity, 3 samples w/40-50% cellularity, M/E: 0.5
Decreased trilineage hematopoesis w/nl maturation
FISH: no abnormalities in 5p15.2, 5q31, 7q31
TERT, TERC, DKC1, TINF2 gene: pending
Final Dx: Dyskeratosis Congenita

Problem Representation: 13F w/skin pigmentation, nail dystrophy, thrombocytopenia p/w epistaxis. Found to be pancytopenic.

Teaching Points (@gabifpucci):

- **Approach to etiology of epistaxis:** predisposition (hereditary-e.g. hemophilia), diseases that increase fragility of the vessels (vit C deficiency), cancers (leukemia), infections, trauma (e.g.inhalation-cocaine use).
- **Investigate important associated factors:** other types of bleeding, systemic symptoms, time course (acute, subacute, intermittent), other skin lesions, headache, meningeal signs
- **Approach to any bleeding:** primary or secondary problems with hemostasis or blood vessel problems
  - Platelet problem: low platelets X increased sequestration X malfunctioning (e.g. vWD) X destructive process
  - Coagulation factor problem: check APT and PT
  - Vessel problem (decreased integrity): vitamin deficiency
- **Approach to a hypoproliferative process** (low platelets + RBC + WBC) -> look at vitamins levels + peripheral smear (destructive process X leukemia + lead deficiency + B12/Folic acid deficiency) -> Bone marrow biopsy
- **Approach to low platelets**: sequestration: splenomegaly, destruction: elevation of LDH, bone marrow failure: leukemia, aplastic anemia, myelodysplastic syndrome, PNH, granulomatous diseases (TB/fungal infections), storage diseases (especially in children), medications, vitamin deficiency (B12, folate acid), autoimmune diseases, infections (HIV, hepatitis, CMV, EBV)