A young patient with progressive LR-MDS

May Basood & Moshe Mittelman

Tel Aviv Sourasky Medical Center
Patient SM: History (I)

- A 41 yo, male, engineer
- 3/2019:
  - Fatigue
  - Macrocytic anemia (Hb 9.3)
- 2017:
  - Asymptomatic
  - Anemia (Hb 11.0)
- PMH:
  - Pericarditis
  - Fatty liver
  - Smoking (20 pack yr)
Patient SM: Family History

- Parents:
  - Cousins

- Father
  - Ischemic heart disease
  - COPD

- Siblings:
  - Brother died in childhood (neuroblastoma)
  - Two healthy: brother and sister
Patient SM: Workup (2019) - I

- **PE:** Unremarkable
- **CBC:**
  - Hb 8.8, MCV 106,
  - WBC 4.4, ANC 3.0, PLT 343
- **Chemistry:**
  - Normal (except: GGT 203u)
- **Anemia workup** - negative
- Diseases **excluded:**
  - Collagen vascular; Infections;
  - Systemic inflammatory
- **Imaging:** Fatty liver; Spleen (15 cm)
Patient SM: Workup (2019) - II

- **Bone Marrow:**
  - Hypercellular (80%)
  - **Erythroid:**
    - Mild hyperplasia
    - Dyserythropoiesis
  - Myeloid: Blasts 3%
  - **Megakaryocytes:**
    - Increased numbers
    - Slightly atypical
    - Ringed *sideroblasts*

- **Cytogenetics:** 46XY
- **Genetics:** SF3B1 mutation
Patient SM: Course (2019-20)

- **Dg:**
  - MDS – RARS - Lower Risk
- **Hb decline**
  - 9.3 – 8.8 – 8.3 – 7.8
- **RBC transfusions**
  - 1-2/4-6 wk
- **Erythropoietin**
  - Eprex 40ku – 80ku/wk
- **No response**
Patient SM: Summary

- A 41 yo male
- RARS - Lower risk MDS
- 46XY
- SF3B mutation
- Hb decline
- RBC transfusion dependent
- ESA resistant
- Sister – matched donor
Patient SM: Questions

- Allo- SCT?
  - Now? Later?
  - Pre-Allo-SCT treatment? (Aza?)

- Experimental treatment?
  - Ruxodustat?
  - Luspatercept?
  - Other?

- Azacitidine?
- Azacitidine + Venetoclax?