



MDS Case 1

BZ

- 68 yo male patient retired mechanic referred for symptomatic anemia x 6 months
- **PMH** stage 1 A colon cancer resected without adjuvant therapy 5 years ago; hyperlipidemia
- **Meds:** atorvastatin
- **SOC:** Married with 3 children. No smoking or ETOH. Father died of AML aged 80.
- **Labs:** Hgb 8.0 g/dL (transfused), WBC $3.0 \times 10^9/L$, ANC $1.8 \times 10^9/L$, Plt $120 \times 10^9/L$, MCV 110fl, RDW 16%
- Cytopenias have been progressive over 5 years, but TD x 6 months
- EPO level is 450 IU/L
- Transfused 2 units of PRBCs every 2-3 weeks- has received in total 18 units of PRBCs, ferritin 1300 ug/L, Iron saturation is 48%.

BZ

- Used to walk 5 km/day, but mainly now sedentary due to dyspnea on exertion
- Physical exam unremarkable, mildly overweight
- All reversible cause of anemia have been excluded
- Blood film- oval macrocytes and occ hypogranular neutrophils
- BM : hypercellular, mild dyserythropoiesis and dysmegakaryopoiesis (5-6%)
- Karyotype: del Y
- NGS: SRSF2 mutation (VAF 20%) and IDH1 mutation (VAF 10%)
- Dx is CCUS

Points to Address

1. What is his risk of developing an overt MN in the next 5-10 years?
2. How would you treat him? Is there a role for hypomethylating agents?
3. When would you consider allogeneic stem cell transplant?
4. How often would you repeat a bone marrow to monitor?
5. What if the patient had only a single DNMT3A mutation at VAF of 18%? How would you manage him?



MDS Case 2

UW Case

- **66 year old male with pancytopenia**

- **Presentation:**

- Pancytopenia x 10+ years
- Pneumonia x 2
- Transfusions: 10 PRBCs, 1 platelet
- Splenomegaly (15.6 cm) & lymphadenopathy (~1.5 cm)
- Sweet syndrome involving skin and pleural effusions in 2019 treated with steroids x 5 months
 - Intermittent chest discomfort
 - Subcutaneous skin nodules on legs of unclear etiology
 - Chronically elevated ESR and CRP

- **Other Medical History:**

- Mild learning disability
- Grave's disease s/p RAI in 2009

11.4 ▼	◆ HEMOGLOBIN (UWH) Ref Range: 13.6 - 17.2 g/dL 8/18/2023 10:45
1.5 ▼	● WHITE CELL COUNT (UWH) Ref Range: 3.8 - 10.5 K/uL 8/18/2023 10:45
46 ▼	× PLATELET COUNT (UWH) Ref Range: 160 - 370 K/uL 8/18/2023 10:45
340 ▼	● ABSOLUTE NEUTROPHILS (UWH) Ref Range: 1,700 - 7,500 /uL 8/18/2023 10:45

10.4 ▲	● C REACTIVE PROTEIN Ref Range: 0.0 - 1.0 mg/dL 6/26/2023 07:06
55 ▲	◆ ESR (UWH) Ref Range: 0 - 15 mm/hr 6/26/2023 07:06

UW Case

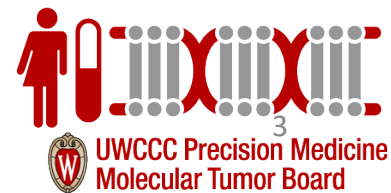
- **Peripheral smear:**
 - No morphologic abnormalities
- **Bone marrow biopsy and aspirate:**
 - Variably cellular marrow (30-50%) with morphologically unremarkable trilineage hematopoiesis, 3% blasts
- **Somatic genetics:**
 - Karyotype: 46,XY, r(7)(p11.1q34), t(14;21)(q13q22.3)[2]/46,XY t(14;21)(q13q22.3)?c[18]
 - Myeloid malignancy panel:
 - *IDH1* (c.394C>A, p.Arg132Ser, VAF 42%)
 - *DNMT3A* (c.2084T>C, p.Ile695Thr, VAF 27%); VUS
- **Germline genetics:**
 - 3q22.1 Interstitial Duplication (7.63Mb, including 66 genes, *GATA2* and *MBD4*)
 - t(14;21) likely constitutional
- **Diagnosis: CCUS**

UW Case

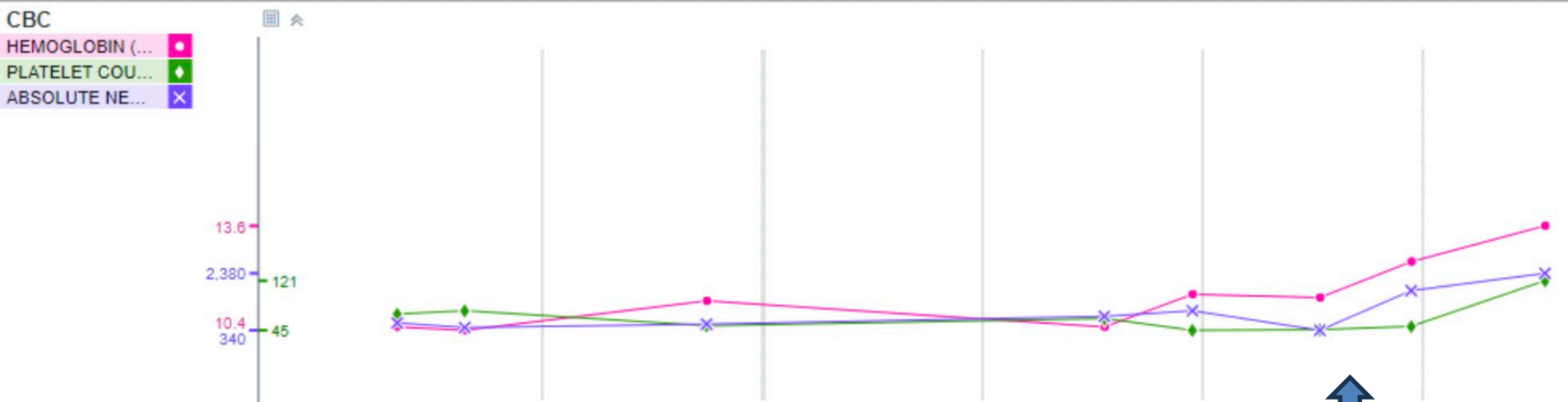
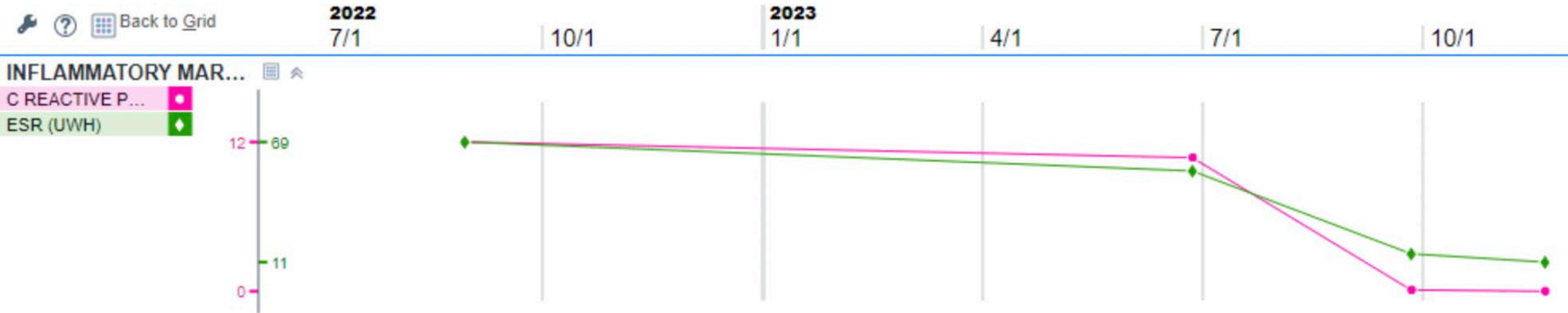
- **Clinical Trial Availability:**

- **NCT 05030441:** A Pilot Study of Ivosidenib for Patients with Clonal Cytopenia of Undetermined Significance and Mutations in *IDH1*
- **Multicenter, Open label, Decentralized, 500 mg/day**
- **PI:** Kelly Bolton, MD, PhD, Washington University
- **Primary endpoint:** rate of improvement in hematologic parameters

Abstract No. 3253 | **Presentation Type:** Poster
Session: 637. Myelodysplastic Syndromes Clinical and Epidemiological: Poster II
Sunday, December 10, 2023, 6:00 PM-8:00 PM



UW Case



Ivosidenib start

UW Case

Baseline

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~3 months

13.6 ◆	◆ HEMOGLOBIN (UWH) Ref Range: 13.6 - 17.2 g/dL 11/20/2023 10:28
4.1 ●	● WHITE CELL COUNT (UWH) Ref Range: 3.8 - 10.5 K/uL 11/20/2023 10:28
121 ▼	× PLATELET COUNT (UWH) Ref Range: 160 - 370 K/uL 11/20/2023 10:28
2,380 ●	● ABSOLUTE NEUTROPHILS (UWH) Ref Range: 1,700 - 7,500 /uL 11/20/2023 10:28

11 ◆	◆ ESR (UWH) Ref Range: 0 - 15 mm/hr 11/20/2023 10:28
0.2 ●	● C REACTIVE PROTEIN Ref Range: 0.0 - 1.0 mg/dL 11/20/2023 10:28

UW Case

- **Drug Tolerance:**
 - **Subjective Improvements in:**
 - Fatigue
 - Chest pains (3x per week to 1-2 per month)
 - Skin nodules (no more)
 - **Potential Side Effects:**
 - Self limiting rash
 - Weight gain (14 lbs)

