MAYO CLINIC

# CASE PRESENTATION

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# Identification

Age: 65

Gender: Male

Allergies: NKDA

## Past Medical History

- Hypertension
- CKD stage II
- Former smoker

| | |
|---|---|
| Chief complaint | Shortness of breath |
| HPI | • 1 week of cough<br>• Progressed to profound shortness of breath now at rest and orthopnea<br>• No changes to medications |
| Physical exam | • Vitals: HR 73, RR 24 BP 184/108 O2 sat 90%<br>• Excessive work of breathing and crackles bilaterally<br>• No edema in the lower extremities |

# LABS

| CBC | |
|---|---|
| Hemoglobin | 7.1 g/dL |
| Hematocrit | 24.4 % |
| MCV | 67 fL |
| RDW | 19 % |
| WBC | 10 x $10^9$/L |
| Platelet count | 270 x $10^9$/L |
| **Differential** | **x$10^9$/L** |
| Neutrophils | 7.8 |
| Lymphocytes | 1.5 |
| Monocytes | 0.42 |
| Eosinophils | 0.09 |
| Basophils | 0.08 |

| Chemistry | |
|---|---|
| Sodium | 133 mmol/L |
| Potassium | 5.8 mmol/L |
| Bicarbonate | 9 mmol/L |
| Anion gap | 20 |
| Creatinine | 9.74 mg/dL |
| BUN | 102 mg/dL |
| Calcium | 7.3 mg/dL |
| Phosphorus | 9.9 mg/dL |
| Osmolality | 324 mOsm/kg |

# IMAGING

- Chest X ray:
  - Small right pleural effusion
  - Perihilar and lower lobe opacities likely reflecting edema
- US kidneys:
  - Increased renal parenchymal echogenicity bilaterally --> CKD
  - No hydronephrosis

# WORKING DIAGNOSES

- Acute hypoxemic respiratory failure
- Hypertensive emergency + Pulmonary edema
- AKI on CKD
- Metabolic acidosis
- Hyperkalemia
- Microcytic Anemia

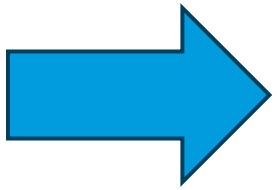
# PLAN

- Patient is started on BiPAP and admitted to the ICU and started on emergent hemodialysis
- Meanwhile they completed the work up for his worsening kidney failure and pulmonary edema

# LATER THAT DAY...

| Initial CBC | |
|---|---|
| Hemoglobin | 7.1 g/dL |
| Hematocrit | 24.4 % |
| MCV | 67 fL |
| RDW | 19 % |
| WBC | 10 x $10^9$/L |
| Platelet count | 270 x $10^9$/L |



| CBC 12 hrs later | |
|---|---|
| Hemoglobin | 5.6 g/dL |
| Hematocrit | 18.3 % |
| MCV | 64 fL |
| RDW | 19 % |
| WBC | 6.2 x $10^9$/L |
| Platelet count | 189 x $10^9$/L |

Confirmed

5.7 g/dL

-No obvious signs of bleeding
-Transfused 2 units of PRBCs

# IMAGING

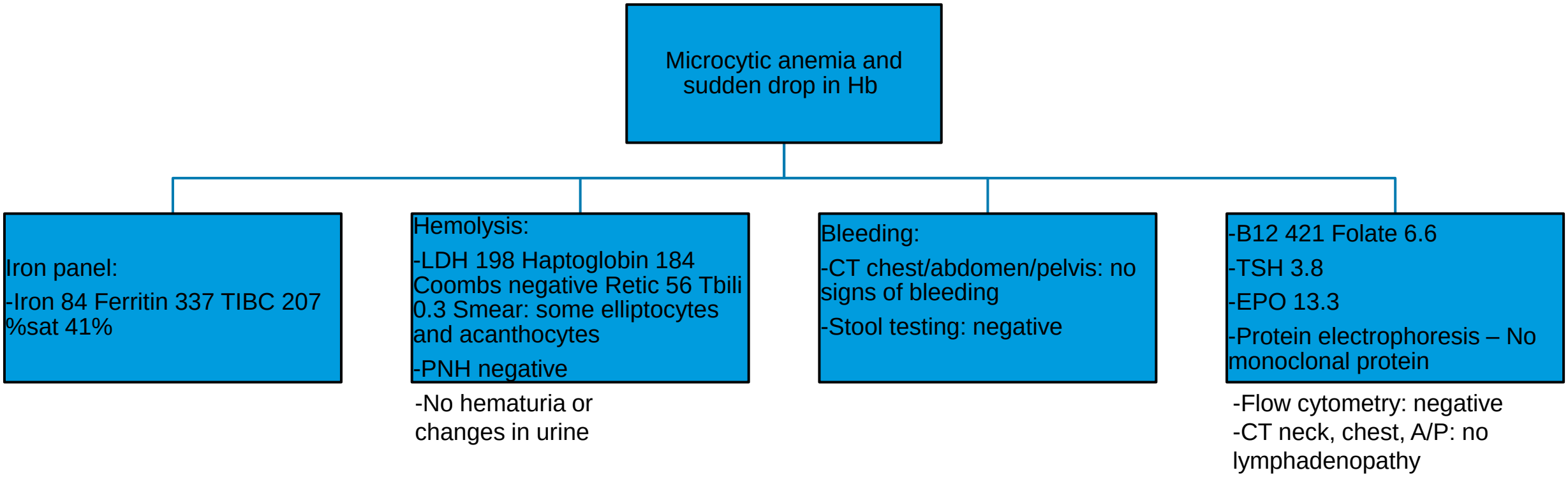
- CT chest abdomen and pelvis:
  - Mild splenomegaly – 16cm
  - 2 cm adrenal adenoma

Anemia evaluation

Hematology consult

# ANEMIA EVALUATION



# ADDITIONAL TESTING

- Kidney biopsy:
  - Diffuse global and focal segmental glomerulosclerosis. Interstitial fibrosis and tubular atrophy, severe, associated with dense interstitial inflammation. Scattered eosinophils.
  - Arteriolar hyalinosis, severe diffuse
  - Overall, the biopsy shows severe arterionephrosclerotic changes likely secondary to hypertension
- Bone marrow biopsy
  - Hypercellular marrow with trilineage hematopoiesis
  - No overt dysplasia is noted in all three lineage hematopoietic elements. Flow cytometry analysis did not reveal abnormal B-cell population or increased blasts. Immunostains for CD34 and CD117 do not reveal any increment of blasts.
  - Erythropoiesis is quantitatively normal. Erythropoiesis is qualitatively megaloblastoid.
  - Hemoglobinization is impaired. Granulopoiesis is slightly increased with normal maturation. Lymphocytes are normal. Plasma cells are normal. Megakaryocytes are normal in appearance. Foreign cells are not present. Myelofibrosis is absent.

# ANEMIA - MOST LIKELY MULTIFACTORIAL

- 1. Anemia of CKD (he was not on ESA)
- 2. Possible hemoglobinopathy
- 3. Sequestration from splenomegaly

# TREATMENT

- Aggressive hypertension management
- Dialysis, high dose steroids, sodium bicarb, ESA
- B12 and folic acid
- Significant improvement of creatinine, BP, breathing and hemoglobin levels

# 3 WEEKS LATER

- Hemoglobin electrophoresis:
  - No electrophoretic evidence of abnormal hemoglobin or beta thalassemia.
- Alpha globin gene sequencing:
  - Positive for alpha thalassemia mutation in alpha-2 globin
- NGS panel from bone marrow biopsy:
  - ASXL1 mutation: c.1782C>A; p.Cys594* (VAF37%)
  - Mutation effect:  The nonsense p.Cys594* mutation results in premature protein truncation before the highly conserved PHD region and is considered inactivating.  Truncating mutations have been shown to result in decreased to absent expression of the wild-type ASXL1 protein and are capable of inducing myelodysplastic syndrome-like disease in mouse models.

# EVENTUALLY…

- The patient underwent renal transplant – currently on mycophenolate mofetil and tacrolimus
- He is currently asymptomatic with good kidney function, hemoglobin baseline 13 g/dL
- He was referred to our CHIP clinic for evaluation where he is being followed every 6 months

# THANK YOU