Through Thick and Thin - Red Blood Cell Disorders

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MUD Meeting 2018
New York City, NY

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Case Study #1

A 42 year old woman has a physical exam in November 2016 at which time a CBC is drawn. The results include a Hgb of 11.5 g/dl (normal 12-16 g/dl), a Hct of 32% (normal 37-47%) and an MCV of 72 (normal 80-100).

- What are the possible causes of these findings?
- What additional information would you want?

Case Study #2

A 74 year old man sees his doctor regularly for follow up. He is known to have a history of hypertension with poor control, rated +50.

His last labs in the APS include a BUN of 30 mg/dl (normal 8-25 mg/dl), a creatinine of 1.8 mg/dl (normal 0.5-1.5 mg/dl) and a CBC with a Hgb of 12.3 g/dl (13-18 g/dl), Hct of 34% (normal 45-52%) and MCV 88 (normal 80-100).

- What is the most likely cause of his anemia?
- Can we offer?

Important Terms

- Too few RBCs – Anemia
- Too many RBCs – Polycythemia
- Anemia can be further divided by size of cells: microcytic, normocytic or macrocytic
- Anemia also can be subdivided by cause: Not enough being made (bone marrow problem), too many being destroyed (hemolysis) or due to blood loss
- Polycythemia: Can be primary (Polycythemia Vera) or secondary (due to testosterone, smoking and others)

Size of Red Blood Cells

- The key value on a CBC report that classifies the cell size is Mean Corpuscular Volume (MCV)
- MCV measures the average volume of Red Blood Cells in a specimen
- Macrocytosis is defined as an MCV > 100 and Microcytosis < 80 – these cut-offs can vary slightly from one lab to another
- Other parameters of the CBC report, such as MCH and MCHC are not typically as helpful in classifying anemias but can be useful in certain specific conditions

Microcytic Anemia

Iron deficiency

- Very important to see an evaluation for the cause
- Could be due to inadequate dietary intake, blood loss due to heavier menses or a more ominous cause such as occult GI bleeding from a malignancy

Thalassemia

- This is a hereditary condition which is more common in individuals with certain regional ancestry.
- The trick to recognizing this is that the MCV is usually low out of proportion to the degree of Hgb/Hct reduction, and the RBC count is almost always normal or even slightly elevated

Normocytic Anemia

Anemia of chronic disease

- This is the most common form of normocytic anemia and the 2nd most common form of anemia after iron deficiency (American Family Physician, November 15, 2000).
- It is related to decreased bone marrow and shortened RBC survival
- Can be associated with inflammatory conditions, infections, neoplasms and various systemic diseases.
- Normocytic anemia is also common in chronic renal insufficiency and certain endocrine disorders such as hypothyroidism or pituitary failure.

Macrocytic Anemia

Megaloblastic

- B12 deficiency
- Folate deficiency
- Certain chemotherapeutic agents

Hemolytic anemia

- Increased reticulocytes
- 20% larger than normal RBCs

Macrocytosis without anemia

- Alcohol
- Liver disease
- Hypothyroidism

Considerations

If Microcytic:

Potential Cause of iron deficiency

- Dietary
- GI blood loss
- GU blood loss (? Menstrual related)

If Normocytic:

- Is there a relevant ‘chronic disease’ present

If Macrocytic:

- B12/Folate levels and replacement
- Alcohol use (may not be anemic)

Case Study # 1

A 44 year old woman has a physical exam in November 2016 at which time a CBC is drawn. The results include a Hgb of 11.5 g/dl (normal 12-16 g/dl), a Hct of 32% (normal 37-47%) and an MCV of 65 (normal 80-100).

- What are the possible causes of these findings?
- What additional information would you want?

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A 74 year old man sees his doctor regularly for follow up. He is known to have a history of hypertension with poor control, rated +50.

His last labs in the APS include a BUN of 30 mg/dl (normal 8-25 mg/dl), a creatinine of 1.8 mg/dl (normal 0.5-1.5 mg/dl) and a CBC with a Hgb of 12.3 g/dl (normal13-18 g/dl), Hct of 34% (normal 45-52%) and MCV 88 (normal 80-100).

- What is the most likely cause of his anemia?
- Can we offer?

Case Study #3

A 42 year old man has a routine check up with his doctor in January of 2017. He had lab tests done which included a CBC: WBC 4.7 (normal 4.0-10.5), Hgb 18.7 g/dl (normal13-18 g/dl), Hct 56.2%(normal 45-52%) and platelet count 389,000 (normal 150-400). MCV was normal.

Looking further in the APS you see that 5 years ago he was seen in Urgent Care for an acute illness and had a CBC with a WBC of 12, Hgb 14.2 and Hct 44 and normal platelets.

The only other pertinent fact is that he started a new work out program one year ago in order to 'get in better shape'.

- What is the differential diagnosis?
- Is there an increased mortality risk?

Case Study #4

A 73 year old woman is noted to have an elevated Hgb and Hct in 2006. At that time a routine CBC was done with a Hgb of 17.5 g/dl (normal 12-16 g/dl) and Hct of 56% (normal 37-47%). The rest of the CBC was normal.

Further work up was done and her jak2 was (+) and erythropoietin level low. Phlebotomy was started.

Over the next 4 years CBC results showed normal to slightly low Hgb and Hct with regular phlebotomy. Then in 2011 her Hgb was fount to be 18.2 and Hct 58.3. She was started on Hydroxyurea and subsequent CBCs have been favorable with Hct < 50%

- What is the most likely diagnosis?
- Can we make an offer?

Primary Polycythemia

- Also known as P. Vera
- Relatively rare condition, 0.6-1.6 per million people (emedicine.medscape.com, 12/2/2016)
- It is a form of Myelodysplastic Disease
- More common in males but does occur in females
- Presence of jak2 mutation
- Complications include thrombosis, hemorrhage and transformation to leukemia
- Higher mortality with younger age of onset (in particular under age <40)
- Treatment options include phlebotomy, Hydroxyurea and cytotoxic drugs

Secondary Polycythemia

- Most often due to reduction in oxygen – COPD, OSA, high altitude
- Special form of non-Primary Polycythemia is called Stress Polycythemia or Gaisböck's Syndrome – related to cigarette smoking or stress
- Diagnostic tests include jak2 mutation, erythropoietin levels and Red Blood Cell Volume

Secondary Polycythemia

"The prognosis of patients with secondary polycythemia is generally related to the prognosis of the underlying disorder. However, the polycythemia itself, when physiologic and not sufficiently extreme to cause significant hyperviscosity, is generally associated with a normal life span. However, emerging evidence suggests that at a minimum, patients with congenital or familial primary polycythemia may have an increased risk of thrombosis".

(emedicine.medscape.com, 8/10/2017)

Considerations

Should always look closely for potential causes of secondary polycythemia before assuming it is P. Vera:

- Any history of OSA
- Living at high altitude
- If COPD, check PFTs
- Use of Testosterone
- Cigarette Smoking

If no clear reason for secondary polycythemia present need to see a work up as P. Vera is a myelodysplastic disorder with significant mortality risk

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