Anemias and Sickle Cell Disease

Introduction to Pathology of Disease

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Thursday, September 25th, 2025

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Case Presentation #1

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A patient comes in to the emergency room…

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- The first step is to collect a patient **history:**
- **Chief Complaint:**
  - A 24 year old African American woman presents with extreme pain in her back and legs.
- **What else would you like to know?**

A patient comes in to the emergency room…

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- **History of Present Illness:**
  - She has had a few episodes like this in the past but this is the most severe pain that she has ever experienced.
  - She is a dental student and finals are next week.
  - Her last menstrual cycle was 3 months ago.

A patient comes in to the emergency room…

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- **Past Medical History:**
  - Wisdom teeth removed at age 17
  - Pneumonia at age 18
- **Family History:**
  - Mother – has similar episodes of pain; Type II diabetes and hypertension
  - Father – hypertension, coronary artery disease, Type II diabetes; had a similar episode of pain and shortness of breath while hiking a mountain in Colorado

A patient comes in to the emergency room…

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- **Review of Systems:**
  - Gastrointestinal – mild abdominal pain
  - Respiratory – shortness of breath and chest pain
  - Genitourinary – burning sensation during urination
  - Remaining systems negative

What other information do we need?

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- **Vital Signs**

	Patient's Vital Signs	Normal Vital Signs
Blood Pressure	110/60	<120/60
Pulse	120	80-100
Oxygen Saturation	94	>95
Temperature	102.1	~98.6

What other information do we need?

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- **Physical Exam**
  - **General:** Tired-looking woman in pain
  - **Skin:** Dry skin and nails
  - **Head and Neck:** Conjunctival pallor and Jaundice/Yellow sclera (white part of the eye)
  - **Cardiovascular:** Tachycardia (fast heart rate)
  - **Other:** Tenderness along her back and left flank
  - Remaining systems are negative

What other information do we need?

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- **Laboratory values**

CBC Component	Patient Results	Normal
White Blood Cells	18,000	3.6 - 11.2 10*9/L
Hemoglobin	6 g/dL	12.9 - 16.5 g/dL
Hematocrit	18%	39.0 - 48.0 %
Platelets	520	150 - 450 10*9/L

Thoughts on what could be going on?

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- Think about a **differential diagnosis.**
- You can use the mnemonic VINDICATE or go by organ system.

VINDICATE		
V	Vascular	
I	Infectious	
N	Neoplastic	
D	Degenerative	
I	Iatrogenic	Intoxication
C	Congenital	
A	Autoimmune	
T	Traumatic	
E	Endocrine	Metabolic

Mnemonic	Possible Diagnoses
Vascular or Hematologic	Anemia
Infectious	Pneumonia, urinary tract infection, chronic/subacute infection
Neoplastic	Cancer – lung, prostate, pancreas, GI, blood
Degenerative	---
Iatrogenic/Intoxication	Substance abuse, medication effect
Congenital	---
Autoimmmune	Rheumatoid arthritis, Multiple sclerosis
Traumatic	Fall leading to pain
Endocrine/Metobolic	Thyroid disease
Psychiatric/Sleep disorder	Insomnia, depression, anxiety, obstructive sleep apnea, periodic leg movements, narcolepsy, chronic fatigue syndrome
Others/Organ System	Liver disease, chronic kidney disease, chronic lung disease, chronic heart disease

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Other Laboratory Values

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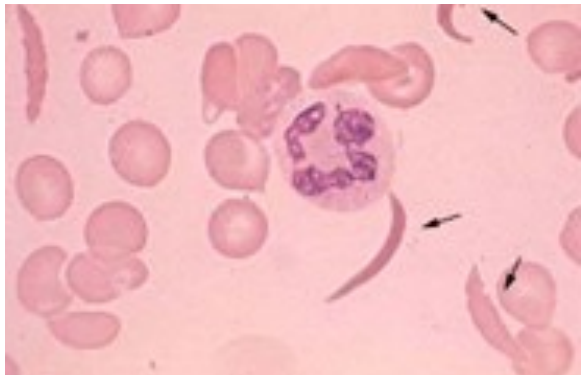
- Urinalysis: numerous bacteria and white blood cells
- Pregnancy Test: Negative
- Peripheral Blood Smear:



- Hemoglobin Electrophoresis: Abnormal hemoglobin, consistent with Hemoglobin S
- **What is going on with the patient?**

Putting It All Together

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- This patient has sickle cell anemia / disease.
- She is experiencing an acute vaso-occlusive pain crisis.
- We confirmed this by several items in our history, physical, and laboratory work-up:
  - Repeated pain (vaso-occlusive) crises over the years
  - Mom and dad have a history of pain crises*
  - Most severe pain crisis ever → precipitated by:
    - Stress – final exams are next week!
    - Urinary tract infection – increased WBC count on CBC; bacteria and WBCs on urinalysis
  - CBC showed decreased hemoglobin and hematocrit → this is anemia
  - Peripheral blood smear showed sickled cells
  - Hemoglobin Electrophoresis showed Hemoglobin S

How can we help her?

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- Pain medicine for the acute vaso-occlusive crisis
- RBC transfusion for the anemia
- Antibiotics for the urinary tract infection
- Hydroxyurea to help increase a different type of hemoglobin in the blood that does not lead to sickle cells
- A doctor’s note to postpone those exams to later date!

Resolution

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- The patient's pain subsides (to her baseline pain level, unfortunately)
- Hemoglobin returns to her baseline of 9 g/dL (still low but typical for sickle cell patients)
- Repeat urinalysis reveals no evidence of bacteria or white blood cells
- She is able to leave the hospital after three days

*What about the family history?

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- **Family History:**
  - Mother – has similar episodes of pain; Type II diabetes and hypertension
  - Father – hypertension, coronary artery disease, Type II diabetes; had a similar episode of pain and shortness of breath while hiking a mountain in Colorado
- What's going on here?
  - Mother has sickle cell disease
  - Father has sickle cell trait

Case Presentation #2

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A patient comes in to the emergency room…

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- The first step is to collect a patient **history:**
- **Chief Complaint:**
  - A 24 year old African American woman presents with shortness of breath and fatigue
- **What else would you like to know?**

A patient comes in to the emergency room…

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- **History of Present Illness:**
  - She started feeling short of breath approximately 2 weeks ago with exercise and now has trouble walking up the stairs. Gets dizzy upon standing and almost passed out yesterday.
  - She is currently on her menstrual cycle, which has lasted for almost 3 weeks.
  - She is a dental student and finals are next week.
  - Has been chewing on ice lately

A patient comes in to the emergency room…

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- **Past Medical History:**
  - Wisdom teeth removed at age 17
- **Family History:**
  - Mother – alive and healthy
  - Father – alive and healthy

A patient comes in to the emergency room…

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- **Review of Systems:**
  - Respiratory – shortness of breath and mild chest pain
  - Genitourinary – no issues urinating
  - Skin/integumentary – dry and cracked
  - Remaining systems are negative

What other information do we need?

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- **Vital Signs**

	Patient's Vital Signs	Normal Vital Signs
Blood Pressure	110/60	<120/60
Pulse	120	80-100
Oxygen Saturation	98	>95
Temperature	98.7	~98.6

What other information do we need?

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- **Physical Exam**
  - **General:** Tired-looking woman in no acute distress
  - **Skin:** Dry skin and nails
  - **Head and Neck:** Conjunctival pallor
  - **Cardiovascular:** Tachycardia (fast heart rate)
  - Remaining systems are negative

What other information do we need?

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- **Laboratory values**

CBC Component	Patient Results	Normal
White Blood Cells	9.0	3.6 -11.2 10*9/L
Hemoglobin	6 g/dL	12.9-16.5 g/dL
Hematocrit	18%	39.0 – 48.0%
Mean Corpuscular Volume	71	80-100 fL
Platelets	520	150-450 10*9/L

Thoughts on what could be going on?

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- Think about a **differential diagnosis.**
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Other Laboratory Values

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- Peripheral Blood Smear:

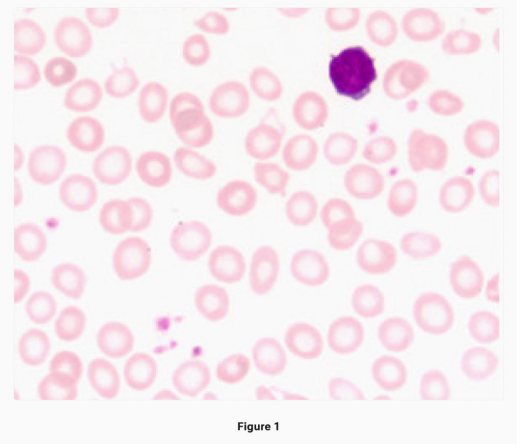

Figure 1

- Iron Studies: Low iron, low ferritin (storage molecule for iron)
- **What is going on with the patient?**

Putting It All Together

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- This patient has iron deficiency anemia from a prolonged menstrual cycle.
- We confirmed this by several items in our history, physical, and laboratory work-up:
  - History of prolonged menstrual cycle
  - Signs and symptoms of anemia: shortness of breath, fatigue, dizziness
  - CBC showed decreased hemoglobin and hematocrit → this is anemia
  - CBC show low mean corpuscular volume (MCV) and Peripheral blood smear showed microcytic red blood cells → microcytic anemia
  - Iron studies shows low iron and low ferritin → this is iron deficiency anemia

How can we help her?

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- Oral or intravenous iron
- RBC transfusion for the anemia
- Oral contraceptives to stop the prolonged menstrual cycle

Resolution

- After RBC transfusion and extended iron use hemoglobin returns to her baseline of 13 g/dL
- She stays on oral contraceptives to prevent prolonged episodes of bleeding