Case Study 4(PKD)

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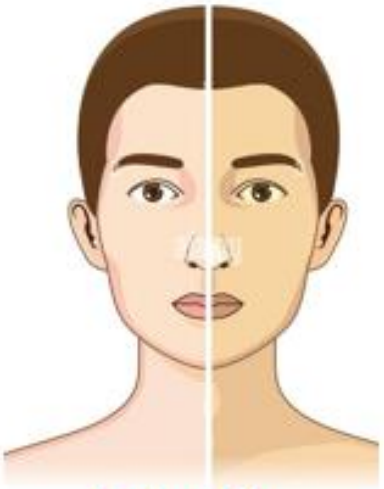
A 22-year-old male presented to the clinic with complaints of **chronic fatigue, shortness of breath on exertion, and pale skin.** He reported having intermittent jaundice since childhood, which worsened during recent viral infections. On examination, he appeared pale with mild scleral icterus, and the spleen was mildly enlarged. Laboratory tests showed **low hemoglobin (9.2 g/dL), elevated indirect bilirubin, increased Lactate dehydrogenase (LDH), and a high reticulocyte count**. **Peripheral blood smear revealed echinocytes (burr cells**), consistent with red blood cell membrane changes seen in pyruvate kinase deficiency. Pyruvate kinase enzyme activity was significantly reduced, confirming the diagnosis of **chronic hemolytic anemia** due to pyruvate kinase deficiency. The patient was managed with folic acid supplementation, monitoring for iron overload, and education on avoiding triggers such as infections. This case highlights how pyruvate kinase deficiency **impairs ATP production** in red blood cells, making them fragile and prone to premature destruction, resulting in hemolytic anemia.

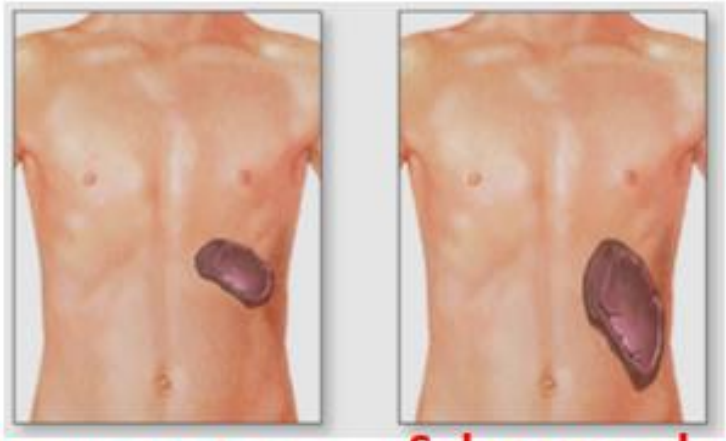
Chronic Fatigue

Shortness of breath



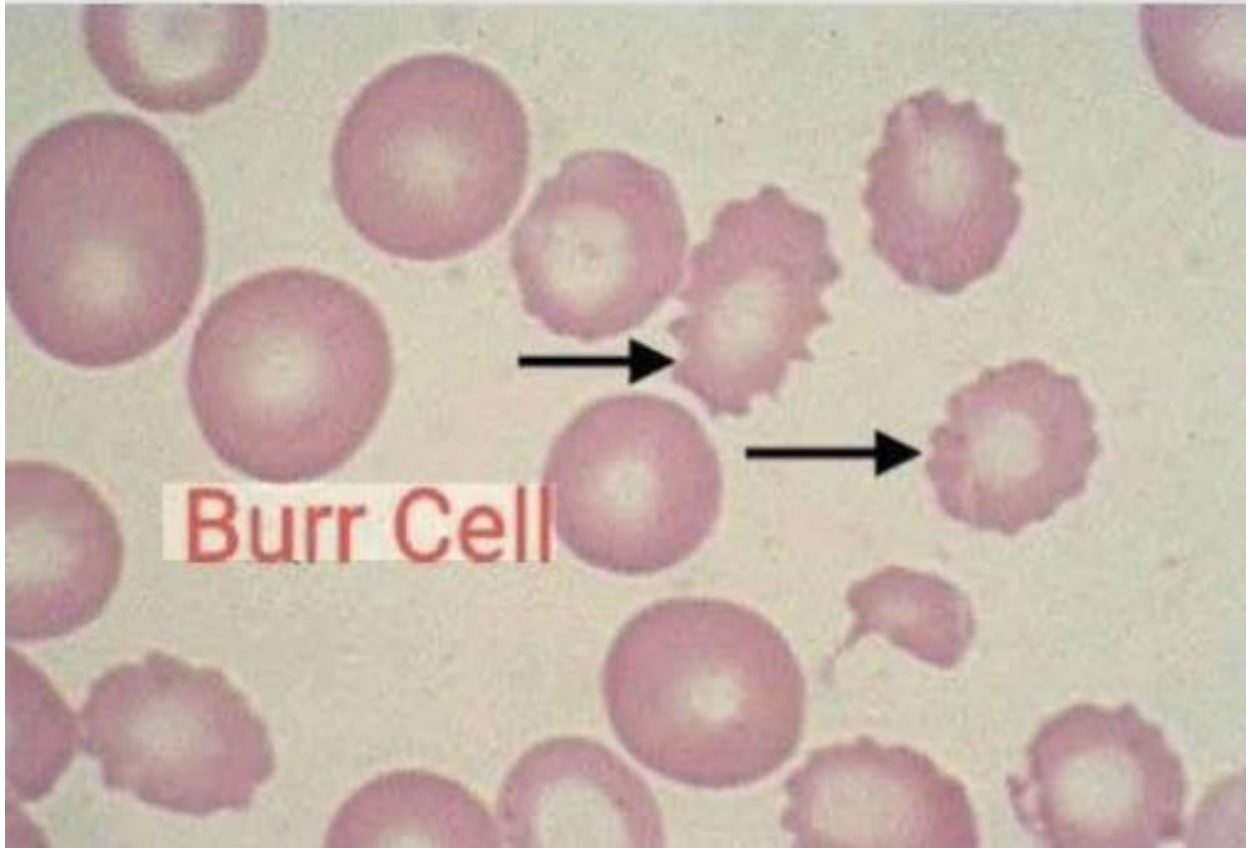
Pale skin

Scleral icterus Normal



Normal spleen Splenomegaly

Peripheral Blood Smear



Sub-Group 1	Question
	1. What were the patient's main symptoms?
	2. What historical clue suggested a long-standing condition?
	3. What physical signs were found during examination?

Sub-Group 2	Question
	4. What was the patient's hemoglobin level?
	5. Which laboratory findings supported hemolysis?
	6. What did the peripheral blood smear reveal?

Sub-Group 3	Question
	7. Which test confirmed the diagnosis of pyruvate kinase deficiency?
	8. What is the primary role of pyruvate kinase in red blood cells?
	9. How does pyruvate kinase deficiency lead to hemolytic anemia?

Sub-Group 4	Question
	10. Why is splenomegaly common in pyruvate kinase deficiency?
	11. Why was folic acid supplementation given to the patient?
	12. What complications should be monitored in patients with PK deficiency?

Sub-Group 5	Question
	13. What factors can worsen hemolysis in PK deficiency?
	14. What is the inheritance pattern of pyruvate kinase deficiency?
	15. How does pyruvate kinase deficiency differ from G6PD deficiency?