Pyruvate Kinase Deficiency

Pyruvate kinase deficiency is an autosomal recessive disease that affects several thousands of people worldwide. Pyruvate kinase catalyzes an ATP-producing step in glycolysis, and when deficient, RBCs are unable to produce the energy required for normal cellular function. This deficiency leads to misshapen cells, known as echinocytes, that are eventually destroyed extravascularly by the spleen and liver, manifesting as normocytic hemolytic anemia. These patients will have elevated 2,3-BPG in their blood.



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Pathophysiology

Autosomal Recessive

Recessive-chocolate

Pyruvate kinase deficiency is an autosomal recessive trait, meaning both parents must pass on one mutated allele each for the offspring to be affected.

Defect in Pyruvate Kinase

Broken Pie-root Kite-ace

Pyruvate kinase is the last enzyme in the glycolysis pathway. It removes a phosphate group from phosphoenolpyruvate to form pyruvate and convert ADP into ATP.

Decreased ATP

Down-arrow ATP-battery

With a deficiency in pyruvate kinase, less ATP is created from glycolysis. Recall that mature RBCs lack mitochondria and thus are solely dependent on glycolysis for ATP production. This deficiency leads to a chronic lack of ATP within the RBCs and interruption of energy-requiring processes.

Extravascular Hemolysis

Extravascular Hemolysis-RBCs

These patients produce inadequate intracellular ATP necessary for plasma membrane Na+/K+ pump functioning, so cells lose water and electrolytes; the subsequent dehydration damages the cell membrane and forms misshapen RBCs, known as echinocytes. These abnormally appearing RBCs are hemolyzed outside of the blood vessels in the spleen and liver in a process known as extravascular hemolysis.

Presentation / Symptoms

Hemolytic Anemia in a Newborn

Hemolysing Anemone and Newborn

Pyruvate kinase deficiency may be in the differential diagnosis for newborns with hemolytic anemia. These patients may have elevated bilirubin from excessive heme breakdown.

Diagnosis

RBC Enzyme Assay

RBC and Enzyme writing an Essay

An RBC enzyme assay is a panel that tests for various RBC enzyme deficiencies in patients with either acute or chronic hemolytic anemia and can confirm a pyruvate kinase deficiency. It may also be useful to order a blood smear, CBC with differential, and reticulocyte count to support the diagnosis.

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Echinocytes

Echidna-cell

Echinocytes, also known as burr cells, may appear on a blood smear for a patient with pyruvate kinase deficiency. These cells have a serrated appearance with small uniform projections that result from poor membrane stability due to a chronic lack of ATP.

Burr Cells

Burr Cell

Burr cells are another name for echinocytes. These thorny, misshapen cells may appear on a blood smear for a patient with pyruvate kinase deficiency. Think of a burr that sticks to clothing to picture the shape of these cells.

Small Uniform Projections

Small-sized Uniform with Projections

Burr cells, or echinocytes, have a misshapen cell membrane that looks like it has many spines or thorns along the surface. In fact, "ekhinos" is the Greek word for "sea urchin."

Normocytic Anemia

Normal-sized-cells Anemone

In normocytic anemia, patients are in an anemic state with a mean corpuscular volume (MCV) between 80 and 100 fL and a corrected reticulocyte count greater than 3%.

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