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Rare cause of anemia in newborns often overlooked, research suggests

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Some babies diagnosed with and treated for a bone marrow failure disorder, called Diamond Blackfan Anemia, may actually be affected by a very rare anemia syndrome that has a different disease course and treatment, say scientists from Dana-Farber/Boston Children's Cancer and Blood Disorders Center.

Genetic analysis of DNA from 175 patients believed to have Diamond Blackfan Anemia, identified eight that showed hallmarks of Pearson Marrow Pancreas syndrome, according to research presented at the 55th annual meeting of the American Society of Hematology.

The treatment choices are difficult in both syndromes, but getting the diagnosis correct is crucial, said Suneet Agarwal, MD, PhD, a pediatric hematologist/oncologist at Dana-Farber/Boston Children's. "Some patients with Diamond Blackfan will respond to steroids, but there's no reason to give steroids to someone with Pearson Syndrome—and they could make things worse," he said.

Diagnosing Pearson Marrow Pancreas syndrome (PS) is not simple, but a specific laboratory test can spot a characteristic abnormality in the infant's DNA that carries blueprints for making proteins in the cells' energy-producing mitochondria.

The test "should be performed in the initial genetic evaluation of all patients with congenital anemia," said Agarwal, who is also affiliated with the Manton Center for Orphan Disease Research at Boston

Children's Hospital.

The two disorders are caused by genetic abnormalities that impair production of blood cells by the bone marrow, causing severe anemia usually diagnosed in the first year of life. Diamond Blackfan Anemia affects approximately one in 100,000 infants and can vary widely in its severity. About 50 percent of patients have physical abnormalities affecting different parts of the body.

Because Diamond Blackfan Anemia is typically inherited from parents in an autosomal dominant fashion, with only one parent carrying the abnormal gene, each pregnancy carries a 50 percent risk of resulting in an affected child.

Pearson Marrow Pancreas syndrome is so rare that fewer than 100 patients have been reported in the literature in the past 25 years, said Agarwal. The genetic defect usually occurs sporadically, he explained, so parents can be counseled that there should be little or no risk of passing along the disease in subsequent pregnancies.

Infants with PS also have anemia and growth defects. They are deficient in pancreatic function and can have muscle and neurologic impairments. Agarwal says it isn't always diagnosed in infancy, because the anemia may not be severe and can even improve without treatment. That's because the patient's cells carry a mixture of normal and mutant mitochondrial DNA. Over time, the proportion of mutant mitochondrial DNA in the blood cells may lessen and the anemia becomes less severe.

Both conditions can be treated with bone marrow transplants, he said, but the risk-benefit calculation is different. "Most patients with Diamond Blackfan Anemia require blood transfusions into adulthood. If you're going to do a transplant in a patient with Diamond Blackfan, outcomes are better if you do it early," Agarwal said.

"Because patients with Pearson Syndrome can get over their blood defect as young children, and because bone marrow transplantation does not cure the other problems in their bodies, the decision to proceed with transplant is more difficult," he added.

Provided by Dana-Farber Cancer Institute