# Diagnosis, Monitoring, and Management of Pyruvate Kinase Deficiency in Children

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## Abstract

Pyruvate kinase (PK) deficiency is an important cause of hereditary non-spherocytic hemolytic anemia caused by a defect in the glycolytic pathway in red blood cells. PK deficient erythrocytes have impaired ATP production and resultant difficulty maintaining normal cell integrity and function, leading to mild to severe anemia due to increased extravascular hemolysis and splenic destruction. Sequelae of chronic hemolysis can result in severe and occasionally life-threatening complications such as hepatobiliary disease, iron overload, bone and cardiopulmonary disease, as well as often markedly impaired quality of life. While the mainstay of management of PK deficiency involves supportive care, comprehensive screening recommendations and disease modifying therapies in development are likely to significantly improve the management of patients. Here, we provide a case-based comprehensive review of the diagnostic evaluation, complications, monitoring recommendations and management of PK deficiency in children.

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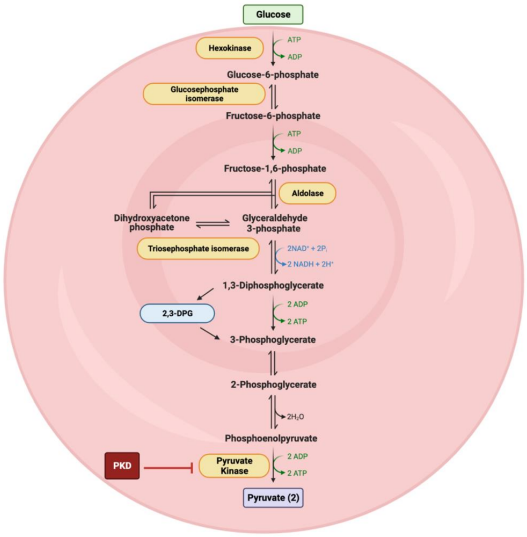


Figure 1.

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PKD Figure 2.docx available at https://authorea.com/users/424368/articles/529466-diagnosis-monitoring-and-management-of-pyruvate-kinase-deficiency-in-children