Pyruvate Kinase Deficiency and Recurrent Splanchnic Venous Thrombosis: A Case Report

Austin Mueller, BS ; Syed-Mohammed Jafri, MD

Henry Ford Health System, Detroit, Michigan

Introduction

- Pyruvate Kinase Deficiency (PKD) is a rare autosomal recessive enzymatic disorder.
- The biochemical consequences of PKD result in red blood cell pyruvate and ATP deficiency, leading to shortened RBC lifespan and chronic, non-spherocytic anemia. [1]
- Additional complications include iron overload, bilirubin gallstones, extramedullary hematopoiesis, and pulmonary hypertension.
- Current treatment approaches are supportive and include transfusion, chelation, and splenectomy.[2]
- Splenectomy usually results in a 1-3 g/dl increase in hemoglobin and may improve the survival of transfused cells.[3]

Case Description

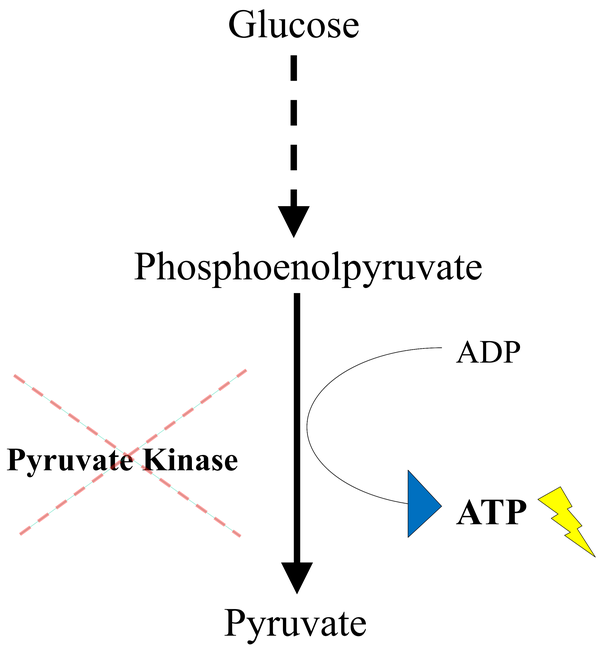
First Thrombotic Episode

- 18 year old male with PKD s/p splenectomy (2001) presenting with RUQ abdominal pain in 2015.
- MRI revealed iron overload and thrombus at the junction of the superior mesenteric vein and portal vein.
- Physical exam was negative for ascites, and the liver was non-enlarged with normal contour.
- Patient was placed on enoxaparin; however, this was shortly discontinued after upper GI bleed from duodenal ulcers.

Second Thrombotic Episode

- 9 months after initial episode, the patient presented with an additional episode of acute RUQ abdominal pain.
- CT scan revealed a new superior mesenteric vein thrombus.
- Patient was initially placed on IV pantoprazole, octreotide, and IV heparin. Patient was bridged to coumadin and has remained hemodynamically stable.

Biochemical Pathway of Glycolysis



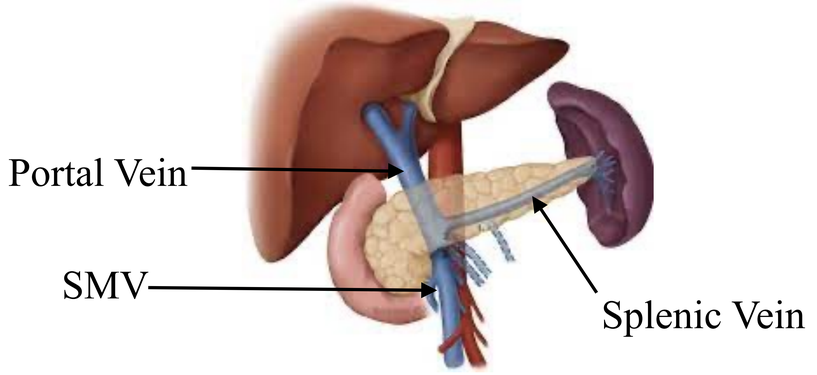
Discussion

- Although splenectomy has shown therapeutic improvement, the risk of post-splenectomy venous thrombotic events is 10% in patients with PKD.[3]

- To our knowledge, only two case reports have detailed recurrent thrombotic events post-splenectomy.
- Patient A received a splenectomy at age 20 with two episodes of portal vein thrombosis at 6 days and 2 years post-splenectomy.[5]
- Patient B received a splenectomy at age 1 with two episodes of pulmonary thromboembolism at 29 and 36 years post-splenectomy.[6]

Why Is Our Case Unique?

- Our patient experienced two recurrent episodes of venous thrombosis involving the superior mesenteric vein (SMV).
- Our patient received a splenectomy at age 5 with thrombotic events at 13 and 14 years post-splenectomy. [7]



Conclusion

- Although splenectomy is a supportive treatment for PKD patients, increased risk for thrombosis has been reported.
- Our case adds to the medical literature by detailing a rare case of PKD with recurrent splanchnic venous thrombosis.
- Although the etiology remains unclear, chronic anticoagulation and regular screening for thrombocytosis is essential to reducing complications and improving prognosis in PKD patients post-splenectomy.

References

1. Al-Samkari H, Van Beers EJ, Kuo KHM, Barcellini W, Bianchi P, Glenthøj A, Del Mar Mañú Pereira M, Van Wijk R, Glader B, Grace RF. The variable manifestations of disease in pyruvate kinase deficiency and their management. Haematologica. 2020 Sep 1;105(9):2229-2239. doi: 10.3324/haematol.2019.240846. PMID: 33054048; PMCID: PMC7556504.
2. Grace RF, Barcellini W. Management of pyruvate kinase deficiency in children and adults. Blood. 2020 Sep 10;136(11):1241-1249. doi: 10.1182/blood.2019000945. PMID: 32702739.
3. Grace RF, Zanella A, Neufeld EJ, et al. Erythrocyte pyruvate kinase deficiency: 2015 status report. *Am J Hematol*. 2015;90(9):825-830. doi:10.1002/ajh.24088.
4. Grace RF, Mark Layton D, Barcellini W. How we manage patients with pyruvate kinase deficiency. Br J Haematol. 2019 Mar;184(5):721-734. doi: 10.1111/bjh.15758. Epub 2019 Jan 25. Erratum in: Br J Haematol. 2019 May;185(4):807. PMID: 30681718.
5. Marcello AP, Vercellati C, Fermo E, Bianchi P, Zaninoni A, Barcellini W, Zanella A. A case of congenital red cell pyruvate kinase deficiency associated with hereditary stomatocytosis. Blood Cells Mol Dis. 2008 Nov-Dec;41(3):261-2. doi: 10.1016/j.bcmd.2008.07.001. Epub 2008 Aug 16. PMID: 18708292.
6. Chou R, DeLoughery TG. Recurrent thromboembolic disease following splenectomy for pyruvate kinase deficiency. Am J Hematol. 2001 Jul;67(3):197-9. doi: 10.1002/ajh.1107. PMID: 11391719.
7. Distal splenorenal and temporary mesocaval shunting at the time of pancreatectomy for cancer: Initial experience from the Medical College of Wisconsin - Scientific Figure on ResearchGate. Available from: https://www.researchgate.net/figure/Anatomy-of-the-superior-mesenteric-vein-SMV-portal-vein-PV-splenic-vein-SV_fig2_234103744 [accessed 6 Apr, 2022]