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Title: CLINICAL AND DEMOGRAPHIC CHARACTERISTICS OF PYRUVATE KINASE DEFICIENCY (PKD) PATIENTS: A COMPREHENSIVE CASE SERIES ANALYSIS

Abstract Type: Publication Only

Topic: Enzymopathies, membranopathies and other anemias

Background:

Pyruvate Kinase Deficiency (PKD) is a rare, autosomal recessive disorder characterized by mutations in the PKLR gene, leading to impaired glycolysis in red blood cells. The enzyme deficiency affects the RBC function and leads to a wide range of clinical manifestations, necessitating personalized management. The prevalence of Pyruvate Kinase Deficiency (PKD) in Saudi Arabia has not been extensively researched. This is particularly notable in a nation where consanguinity is common, and the overall prevalence of congenital hemolytic anemia presents a unique challenge in identifying specific disorders, especially within non-specialized medical facilities.

Aims:

In this paper we are reporting case series to provide an in-depth examination of the clinical and demographic characteristics of PKD patients, shedding light on its heterogeneity.

Methods:

We conducted an extensive retrospective analysis of seven PKD patients of Arab ethnicity who were under our care. Data encompassed demographic information, detailed medical history, clinical features, laboratory results, treatment regimens, and clinical outcomes.

Results:

The group of patients under study consisted of 5 males and 2 females, whose ages ranged from 10 to 38 years. All individuals in this cohort identified as Arab ethnicity.

Regarding their family backgrounds, all patients except one had consanguinity in their families.

In our observed patient group, the onset of symptoms was consistent across the board, manifesting in early ages as either neonatal jaundice or symptomatic anemia.

One patient developed severe hepatic disease with evolution to liver failure followed multiorgan failure and death.

The PK activity was done on all patients, and all were below 19% (range 0.75- 18.2), genetic testing was done for one patient and showed pathogenic variants in PKLR.

The necessity for blood transfusions was universal among all patients, illustrating the severe clinical nature of their condition.

Hemoglobin levels showed considerable variation, ranging from 6.5 to 9 g/dL.

There was no report of myelodysplastic syndrome, leg ulcers or pulmonary hypertension.

The majority of patients, except one, underwent a Splenectomy as part of their treatment strategy and the reasoning for it were to improve baseline anemia, decrease transfusion dependency and to improve patient quality of life.

Following the splenectomy 3 patients had a rise in the hemoglobin level and decrease in the need for transfusion support, while the other patients were not able to discontinue transfusions following the surgery.

Thrombocytosis was evident in 5 of the patients post splenectomy and 1 patient developed venues thromboembolism in form of bilateral pulmonary embolism.

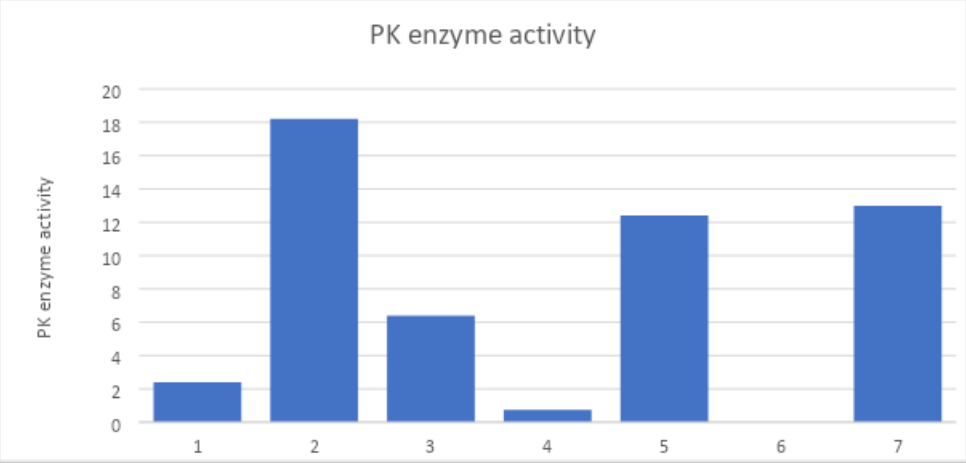
Jaundice was a common clinical and biochemical observation among all patients and was complicated the by presence of gallstone, a frequent complication of PK deficiency and for which cholecystectomy was performed on 6 patients.

Commonly prescribed medications in our cohort included folic acid, iron chelation, prophylactic antibiotics, and aspirin.

Summary/Conclusion:

This study provides valuable insights into the diverse and complex clinical manifestations of Pyruvate Kinase Deficiency (PKD). Our analysis of a cohort comprising both males and females of Arab ethnicity, spanning a wide age range, highlights the hereditary nature of the disorder, as evidenced by consanguinity and family history among patients. The findings emphasize the severe impact of PKD, necessitating universal blood transfusion and often requiring splenectomy to manage anemia and enhance quality of life.

Figure 1: PK enzyme activity.



Keywords: Pyruvate kinase, Pyruvate kinase deficiency