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Title

Clinical and Demographic Characteristics of Pyruvate Kinase Deficiency (PKD) Patients: A Comprehensive Case Series Analysis

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Introduction

Pyruvate Kinase Deficiency (PKD) is an autosomal recessive enzymopathy disorder leading to hemolytic anemia. The disorder is caused by mutations in the PKLR gene, and its prevalence remains largely unknown, particularly in populations like Saudi Arabia where consanguinity is common. This paper provides a detailed examination of a case series of PKD patients to shed light on the clinical heterogeneity of this condition.

Methodolgy

We performed a comprehensive retrospective study of seven PKD patients of Arab ethnicity, focusing on demographics, clinical features, laboratory findings, and treatment outcomes.

Results

Our study involved seven patients of Arab ethnicity with Pyruvate Kinase Deficiency (PKD), comprising 5 males and 2 females, ages ranging from 10 to 38. Family history suggested a hereditary nature of PKD, as all but one patient had consanguinity and five had a family member with PKD. The clinical manifestations of the disease were diverse but universally severe, commonly presenting as neonatal jaundice or symptomatic anemia in early life (Table 1). Blood tests revealed PK activity below 19% in all patients, confirming the disease's severe nature, which necessitated universal blood transfusion (Figure 1). Anemia severity varied, with approximately 67% having mild to moderate anemia and the remaining 33% experiencing severe anemia. Hemoglobin levels also varied widely, ranging from 6.5 to 9 g/dL. Treatment strategies primarily included splenectomy to improve anemia and decrease transfusion dependency. Post-splenectomy, three patients showed improved hemoglobin levels and reduced transfusion need. However, thrombocytosis was common post-surgery, with one patient developing venous thromboembolism. Iron chelation was universally employed to manage iron overload, yet the issue persisted, indicating a need for ongoing management. Jaundice was universally observed and often complicated by gallstones, leading to cholecystectomies in six cases. Lab findings across all patients were consistent, marked by elevated reticulocyte counts, lactate dehydrogenase levels, and indirect hyperbilirubinemia, alongside decreased haptoglobin levels. The common medication regimen included folic acid, iron chelation, prophylactic antibiotics, and aspirin. There were no cases of myelodysplastic syndrome, leg ulcers, or pulmonary hypertension.

Conclusion

Our study reveals a complex picture of PKD, emphasizing the need for personalized treatment and the complications arising from the disease. These insights contribute to a better understanding of PKD, particularly in regions where consanguinity is prevalent.