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Rare Case of Haemolytic Anaemia Due to Pyruvate Kinase Deficiency (PKD)

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Abstract

Pyruvate kinase deficiency (PKD) is a rare autosomal recessive cause of congenital non-spherocytic haemolytic anaemia, resulting from mutations in the PKLR gene. We report a 14-year-old patient presenting with right hypochondrial pain, subicterus, and splenomegaly. Laboratory tests showed normocytic anaemia, reticulocytosis, indirect hyperbilirubinaemia, and reduced pyruvate kinase activity, confirming PKD. Imaging revealed biliary lithiasis and splenomegaly. PKD should be considered in cases of chronic haemolysis with indirect hyperbilirubinaemia. Early diagnosis and multidisciplinary follow-up improve outcomes.

Keywords: Pyruvate kinase deficiency; PKLR gene; haemolytic anaemia; splenomegaly; enzyme deficiency; case report

I. INTRODUCTION

Pyruvate kinase deficiency (PKD) is the most frequent cause of non-spherocytic haemolytic anaemia. It is an autosomal recessive disorder caused by mutations in the PKLR gene located on chromosome 1q22. To date, more than 190 mutations have been identified in this gene [1].

II. Epidemiology

The prevalence of PKD remains imprecise, estimated between 3 and 8 cases per million inhabitants [2]. Because of its rarity, diagnostic challenges, and broad clinical variability, many cases may remain undiagnosed, leading to an underestimation of its true frequency.

Heterozygous carriers are typically asymptomatic, making it difficult to evaluate their prevalence, which is nonetheless estimated between 0.15% and 6% [3,4].

III. Case Report

We report the case of a 14-year-old patient with no significant past medical history, who presented with right hypochondrial pain suggestive of hepatic colic.

Clinical examination revealed a patient in good general condition, with subicterus, moderately icteric conjunctivae, tenderness in the right upper quadrant (without guarding), stage II splenomegaly, and no evidence of ascites, cardiovascular collapse, or lymphadenopathy.

Laboratory findings:

- Haemoglobin: 8.5 g/dL

- MCV: 89 fL

- MCHC: 29.01%

- Platelet count: 377,000/mm³

- Reticulocyte count: 247,260/mm³

- Total bilirubin: 34 mg/L (predominantly indirect: 28.4 mg/L)

- Liver function tests: within normal limits

- Direct Coombs test: negative

- Haemoglobin electrophoresis: normal

- Enzyme assay: confirmed erythrocyte pyruvate kinase deficiency, with markedly reduced enzymatic activity and elevated G6P2/PK ratio

- Genetic testing was not performed.

Abdominal ultrasound showed biliary lithiasis without complications, and moderate, homogeneous splenomegaly.

IV. Discussion

PKD is a rare autosomal recessive disorder of the glycolytic pathway, first described in the early 1960s [5–8]. It leads to congenital, chronic haemolytic anaemia due to impaired energy production in erythrocytes.

The diagnosis should be suspected in patients with chronic haemolysis presenting with jaundice, splenomegaly, indirect hyperbilirubinaemia, and elevated reticulocyte count. Diagnosis is confirmed by enzyme activity testing and, when available, molecular analysis of PKLR gene mutations [6,9].

Management is challenging and often includes supportive measures such as blood transfusions, splenectomy, and, in specialised centres, disease-modifying therapies [10].

V. Conclusion

Pyruvate kinase deficiency is a rare but significant cause of congenital haemolytic anaemia. Due to its chronic course and potential complications, long-term multidisciplinary follow-up is essential to improve patient outcomes.

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