Case Report

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Gilbert Syndrome Presenting as Jaundice in an adolescent Boy : A Case Report

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Abstract

Gilbert Syndrome (GS) is a hereditary condition characterized by intermittent hyperbilirubinemia due to a mutation in the UGT1A1 gene. This case report discusses a 12-year-old male presenting with jaundice and mild abdominal discomfort. Laboratory investigations revealed isolated unconjugated hyperbilirubinemia with normal liver function tests, leading to the suspicion of GS. Genetic testing confirmed a homozygous polymorphism in the UGT1A1 gene. Management included patient education and regular monitoring, with no specific treatment required. This case highlights the importance of considering GS in the differential diagnosis of jaundice in adolescents and the role of genetic testing in its confirmation. Early diagnosis can prevent unnecessary interventions and ensure appropriate management.

Keywords:- Gilbert Syndrome, Adolescent, Jaundice, Hyperbilirubinemia, UGT1A1 Gene

INTRODUCTION

Gilbert Syndrome (GS) is a common hereditary disorder characterized by intermittent episodes of mild jaundice due to unconjugated hyperbilirubinemia. It is caused by a mutation in the UGT1A1 gene, leading to reduced activity of the enzyme uridine diphosphate-glucuronosyltransferase (UGT).[1] This enzyme is crucial for the conjugation and excretion of bilirubin, a byproduct of hemoglobin breakdown. GS is generally benign and often discovered incidentally during routine blood tests that reveal elevated levels of unconjugated bilirubin. Clinical manifestations are typically mild, with jaundice being the most prominent symptom, especially under stress, fasting, or illness. [2]

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GS is prevalent worldwide, affecting approximately 3-12% of the population. The condition is more frequently diagnosed in males, likely due to hormonal influences that affect bilirubin metabolism. It is often identified in adolescence or early adulthood but can remain undiagnosed until later in life. The pathophysiology involves a reduction in the conjugation capacity of bilirubin in the liver, resulting in its accumulation in the bloodstream. [3]

Clinically, GS presents with intermittent jaundice without other systemic symptoms. In children and adolescents, the diagnosis is challenging as jaundice can be attributed to various other conditions. The differential diagnosis includes hemolytic anemia, hepatitis, and other liver disorders. Diagnosis of GS involves ruling out other causes of jaundice through a combination of clinical history, physical examination, and laboratory tests. Genetic testing can confirm the diagnosis by identifying mutations in the UGT1A1 gene. [4]

A notable aspect of GS is the isolated increase in unconjugated bilirubin without evidence of hemolysis or liver dysfunction. This distinction is crucial for management, which generally involves reassurance and monitoring rather than invasive interventions.[5]

CASE REPORT

The patient is a 12-year-old male with a history of intermittent jaundice. He presented to the clinic with a two-week history of worsening jaundice, fatigue, and mild abdominal discomfort. There was no history of fever, weight loss, or changes in appetite. The patient's medical history was unremarkable, with no previous hospitalizations or significant illnesses. He had no family history of liver disease or hemolytic disorders.

On physical examination, the patient appeared well-nourished and alert. Scleral icterus was noted, and jaundice was evident on the skin. There were no signs of hepatosplenomegaly, and the abdomen was soft and non-tender. No other abnormalities were detected on systemic examination.

Test	Result	Normal Range
Total bilirubin	5.8 mg/dL	0.1 - 1.2 mg/dL
Direct bilirubin	0.3 mg/dL	0.0 - 0.3 mg/dL
Hemoglobin	13.8 g/dL	12.0 - 15.5 g/dL
Reticulocyte count	1.8%	0.5 - 2.5%
Liver function tests	Normal	
Blood group	B+	
Coombs test	Negative	Negative
Glucose-6-phosphate dehydrogenase (G6PD) level	Normal	Normal
Hepatitis panel	Negative	Negative

Table 1:- Lab investigations in studied case.

Given the elevated unconjugated bilirubin and normal liver function tests, genetic testing was conducted. The results confirmed a homozygous polymorphism in the promoter region of the UGT1A1 gene, consistent with Gilbert Syndrome.

Management focused on educating the patient and family about the benign nature of the condition. No specific treatment was required, but the patient was advised to avoid fasting and to maintain a healthy diet. Regular follow-ups were scheduled to monitor bilirubin levels and overall health.

DISCUSSION

This case highlights the importance of recognizing Gilbert Syndrome as a cause of jaundice in children and adolescents. GS is often overlooked due to its benign nature and the intermittent presentation of symptoms. The diagnosis in this case was confirmed through genetic testing, which is crucial for differentiating GS from other causes of hyperbilirubinemia.[6]

Similar cases in the literature have emphasized the variability in presentation and the importance of genetic confirmation. For instance, a study by Bosma et al. detailed the molecular diagnosis of GS, underscoring the role of UGT1A1 gene mutations in the condition.[7] Another report by Owens et al. discussed the clinical management of GS in adolescents, highlighting the need for appropriate counselling and follow-up.[8] Additionally, Kadakol et al. described the genetic

and clinical aspects of GS, providing insights into the management of affected individuals. [9]

An important discussion point is the management strategy for GS, which primarily involves reassurance and monitoring. In this case, the patient was educated about the condition, and unnecessary interventions were avoided. The use of genetic testing was pivotal in confirming the diagnosis and guiding the management plan.[10]

CONCLUSION

Gilbert Syndrome should be considered in the differential diagnosis of jaundice in adolescents. Genetic testing plays a crucial role in confirming the diagnosis and distinguishing GS from other more severe conditions. Management primarily involves reassurance and regular monitoring, emphasizing the benign nature of the disorder.

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