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A Severe Hyperbilirubenmia in Gilbert Syndrome

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Abstract

Bilirubin is a waste product produced by the breakdown of red blood cells. Gilbert Syndrome is a genetic disorder that affects the liver's ability to process bilirubin (1). Gilbert syndrome consists of periodic episodes of mild jaundice, which can cause symptoms such as fatigue, abdominal pain, and dark urine (2). Gilbert syndrome is often misdiagnosed for a more serious liver disorder, however, it is a benign syndrome.

Most cases of Gilbert syndrome consist of hyperbilirubinemia. Typically bilirubin levels in Gilbert Syndrome is less than three, with rare instances of it being above 3; however, it typically does not surpass 6 mg/dL. This case study describes a unique clinical presentation of a patient with Gilbert syndrome. This case is unique because the patient consistently has hyperbilirubinemia and it is at levels above 20 mg/dL. Additionally this case is unique in that the patient did not have any abdominal pain or signs of hepatic disease. Given the unique presentation of this disease, it is important to share our approach with others.

Keywords: Bilirubin; Gilbert Syndrome; Hyperbilirubenmia

Introduction

Gilbert Syndrome is a genetic disorder that affects the liver's ability to process bilirubin, a waste product produced by the breakdown of red blood cells [1]. It is characterized by periodic episodes of mild jaundice, which can cause symptoms such as fatigue, abdominal pain, and dark urine [2]. While Gilbert Syndrome is generally a benign condition, it is sometimes misdiagnosed as a more serious liver disorder, making an accurate diagnosis and proper management important. This case study aims to describe the clinical presentation, diagnosis, and management of Gilbert Syndrome in a 22-year-old male.

Case Report

A 22-year-old male with a history of Gilbert Syndrome presented to the emergency department per his primary care physician's recommendation following recent lab work that demonstrated a total bilirubin level of 10 mg/dL. On presentation, the patient had no complaints.

In the emergency department, the physical exam demonstrated an alert and oriented, healthy patient in no acute distress with jaundice. Additionally, the patient's oral mucosa appeared to have decreased moisture. CBC with auto differential, BMP, Hepatic Function Panel, Urinalysis, Lipase, Prime-INR, APTT, Hepatitis Panel-Acute, and plasma

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lactic acid was ordered. CBC was normal except for an MCH of 34.4 pg, an MCHC of 36.7 g/dL, and a monocyte absolute of 0.80 (10*3/uL). BMP was normal except for an anion gap of 3 mmol/L. The Hepatic Function Panel demonstrated a total bilirubin of 13.4 mg/dL and a direct bilirubin of 0.30 MG/DL. There was no bilirubin in the urine. All other laboratory findings were normal. Ultrasound of the right upper quadrant demonstrated mild contraction with mild wall thickening. The patient was given intravenous fluids and scheduled for a follow-up with a gastroenterologist.

During his visit with the gastroenterologist, the patient reported that he has had episodes of elevated bilirubin before, with total bilirubin levels above 20 mg/dL. His most recent elevated number prior to his presentation to the emergency department was 1 year and 3 months prior and had a value of 6 mg/dL. The patient could not attribute his episodes of hyperbilirubinemia to any inciting event. He stated that his skin is always jaundiced; however, severity is dependent on stressors and sun exposure. Patient was counseled on ultraviolet light and its impact on jaundice. Given the patient's stable condition and lack of symptoms, liver enzymes, and bilirubin levels will be continually monitored. The following laboratory tests were ordered during this encounter: Iron, Ferritin, Transferrin, Antinuclear antibodies, Anti-smooth muscle antibodies (IgG), Mitochondrial antibodies (M2), Alpha-1-antitrypsin, Hepatitis A antibody, Hepatitis B surface antibody, ceruloplasmin, and anti-microsomal antibody. Of these tests, ceruloplasmin was abnormal with a value of 18 mg/dL. Hepatitis A antibody was positive and the anti-microsomal antibody was 40 units.

Discussion

Gilbert Syndrome is an autosomal recessive condition that is the result of decreased activity of uridine diphosphate glucuronyl transferase 1A1 (UGT1A1), an enzyme that plays a role in the conjugation of bilirubin [1]. This enzyme's function is typically reduced to 30% of normal [3]. Gilbert syndrome is characterized by unconjugated hyperbilirubinemia and jaundice. Individuals with Gilbert syndrome usually have their first instance of jaundice as teenagers or young adults [2].

Typically, patients with Gilbert Syndrome are asymptomatic. However, when symptoms are present they usually consist of mild intermittent jaundice, abdominal pain, and fatigue. Jaundice can be triggered by hemolysis, stress, physical exertion, febrile illness, fasting, and menses [2]. Hyperbilirubinemia in these patients is usually less than 3 mg/dL. In some instances, such as pathologic conditions, plasma bilirubin concentrations may be higher than 3 mg/dL but typically do not surpass 6 mg/dL [4].

There have been previous cases with severe unconjugated hyperbilirubinemia, such as seen in the patient above. The patients in these case reports have an additional mutation or disorder that contributes to hyperbilirubinemia. Some examples include hemolytic disorders, an additional mutation in UGT1A1, or heterozygous carriers Crigler-Najjar-type structural mutation [5]. There are two cases, one of a patient with severe hyperbilirubinemia secondary to hereditary spherocytosis and another case with an additional mutation in UGT1A1 who had successful treatment with rifampicin 600 mg/day. Rifampicin is assumed to treat hyperbilirubinemia via induction of UGT1A1. In both these cases, the patients had severe symptoms of disabling fatigue and pruritus [6].

Patients with Gilbert Syndrome and severe hyperbilirubinemia should have their bilirubin regularly checked. The development of severe symptoms could be an indication of Rifampicin treatment [6].

Conclusion

This case study illustrates the importance of considering Gilbert Syndrome in the differential diagnosis of jaundice and abdominal pain, even in the absence of other signs of liver disease. Genetic testing can confirm the diagnosis and guide management. With proper management, patients with Gilbert Syndrome can lead healthy, symptom-free lives.

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