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Case Report

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Patient with Gilbert Syndrome A Case Report and Review of the Literature

Walaa Fikry Elbossaty

Department of Biochemistry, Faculty of Sciene, Damietta University, Egypt

***Corresponding author**
Walaa Fikry Elbossaty, Department of Biochemistry, Faculty of Sciene, Damietta University, Egypt

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Abstract

One of the Gilbert syndrome genetic diseases that is characterized by the presence of a genetic mutation in UGT1A1 gene, which is responsible for uridine diphosphate glucuronosyl transferase enzyme, responsible for the binding of bile, and thus the ease of its release from the liver and its non-accumulation in the body.

A middle-aged man suffers from jaundice, although all clinical, laboratory, and imaging tests are normal. Hemolysis was excluded by the fact that the blood image did not include the presence of foreign cells, as well as reticulocyte count, and hemoglobin concentration in their normal rates, so it was excluded that the main cause of high bilirubin was the occurrence of hemolysis. So he was diagnosed with Gilbert's syndrome.

Keywords: Gilbert's syndrome, UGT1A1 gene, Bilirubin, UGT

Introduction

There are two types of bilirubin conjugated (water soluble), unconjugated bilirubin (water insoluble). In order for the liver to get rid of bilirubin, it must be in conjugated form. Uridine diphosphate glucuronosyl transferase (UGT) is an effective role in converting toxic form (un conjugated) bilirubin to non-toxic form (conjugated) bilirubin in a process known as glucuronidation, and therefore easy to handle and excrete through the liver [1].

The gene responsible for expressing that UGT enzyme is called UGT1A1 gene. This gene is located on chromosome 2 long arm at position 37.1 (Fig.1) [2].

It was found that any genetic mutation in UGT1A1 leads to a deficiency of the UGT enzyme responsible for glucuronidation, and thus accumulation of bilirubin especially un-conjugated bilirubin [3].

Review of the Literature

Gilbert's syndrome was first discovered by Augustin Gilbert and Pierre Lereboullet in 1901, where the syndrome was classified as an increase in the amount of unrelated blood yellow and symptoms usually appear in adulthood and adolescence. Incidence of disease in male more than in female [4].

It has been found that the reason for the high incidence of bile is related due to a deficiency in the enzyme uridine diphosphate glucuronosyl transferase and the person responsible for converting the unrelated bilirubin into a link to facilitate handling through the liver and disposal of them in various metabolism processes to prevent their accumulation inside the body and the emergence of the so-called hyperbilirubinemia. Research has shown that the reason for this enzyme deficiency is a genetic mutation in UGT1A1 gene. Gilbert's syndrome has a 3-7% prevalence of the world's population [5].

In Gilbert's syndrome, it was found that the genetic mutation that occurred in UGT1A1 gene at location 71 where the amino acid glycine was replaced to the amino acid arginine and written on this picture Gly71Arg or G71R and hence the effect of this on enzyme function. Studies have found that patients with Gilbert's syndrome have only 30% of the enzyme's function [6].

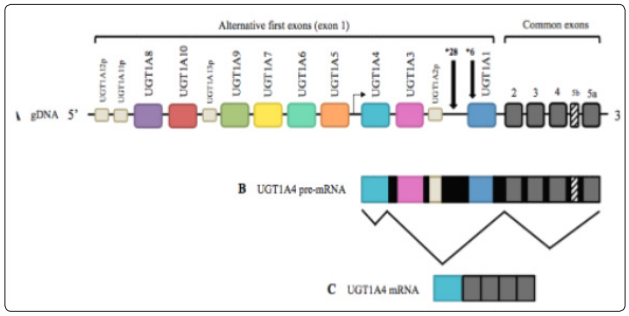

Figure 1: Human UGT1A locus

Case Report

Our patient is a 42-year-old man who has been suffering from a high rate of bile in the blood for a long time, but he is in good health and does not suffer from any diseases.

The case underwent a number of clinical, laboratory and imaging examinations, and all tests confirmed that the man does not suffer from any diseases except for the high rate of bilirubin.

Clinical examinations did not show any symptoms, and imaging tests using ultrasound demonstrated the absence of hepatomegaly or splenomegaly.

The blood picture showed that: hemoglobin concentration is 15 mg / dL, the number of red blood cells is 5.2 mm3, and the percentage of platelets is 345 mm3. No foreign cells appeared, and the Reticulocyte count was normal, direct Coombs test was negative indicating the absence of hemolysis.

Medical analyzes showed that the rate of liver enzymes in their normal levels where the rate of sGOT 22 U / L, the rate of sGPT 28, and albumin 3.4 3.4 gm. / dL, and total bilirubin 2.8 mg / dL, and direct bilirubin 0 .34 mg / dL. No viral hepatitis (HCV Ab and HBsAg). No yellowing in urine or stool, no nausea, vomiting or abdominal pain

Discussion

Through the interpretation of the results and the merging between clinical, laboratory and imaging tests, it was found that the condition of the liver is intact, the enzymes are in their normal range, and the liver performs its normal functions except for the rise of total bilirubin including indirect bilirubin. The presence of hemolysis all of this indicates that the rate of bilirubin, especially indirect, is not high due to a reason for the breakdown of red blood cells, as bilirubin modifies the products of the breakdown of hemoglobin resulting from the breakdown of red blood cells [7].

All these indications confirmed that this condition suffers from Gilbert's syndrome. This can be confirmed by establishing a genetic mutation in the UGT1A1 gene.

Gilbert syndrome is easy to diagnose with qPCR technique for TATA box of the UGT1A1gene [8].

It is also possible to control hyperbilirubinemia through good nutrition and to take phenobarbital when the bilirubin level rises significantly [9].

Conclusion

Gilbert's syndrome is one of the genetic diseases that have no symptoms and does not affect the vitality of the affected person except for the high rate of bilirubin. Therefore, the injured person should enjoy calmness, not panic and panic because this condition has no developments or repercussions on his life. But he should avoid stress and take medications without medical supervision because there are some drugs that affect the high rate of bilirubin in the blood.

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