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Opinion Article

UGT1A1 Gene Plays a Significant Role in Gilbert's Syndrome

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DESCRIPTION

Gilbert Syndrome (GS) is a condition in which the affected person's liver processes bilirubin more slowly than the general population. It has a lot of health benefits. Many people never experience any symptoms. Jaundice (a mild yellowish hue to the skin or whites of the eyes) can develop on occasion.

Gilbert syndrome is caused by a genetic variation in the *UGT1A1* gene, which causes the bilirubin uridine diphosphate glucuronosyltransferase enzyme to be less active. Depending on the type of mutation, it is usually inherited in an autosomal recessive pattern, although it can also be inherited in an autosomal dominant form. Stress, such as exercise, menstruation, or not eating, can cause episodes of jaundice. Higher levels of unconjugated bilirubin in the blood without any additional signs of liver disease or red blood cell disintegration are used to make the diagnosis.

Cause

Gilbert Syndrome is caused by mutations in the *UGT1A1* gene. The gene codes for the enzyme bilirubin uridine diphosphate glucuronosyltransferase (bilirubin-UGT), which is present in liver cells and is responsible for the elimination of bilirubin from the body.

The glucuronidation reaction is carried out by the bilirubin-UGT enzyme. Glucuronic acid is transformed into conjugated bilirubin from unconjugated bilirubin, a yellowish pigment produced when your body breaks down old red blood cells. Bile transports conjugated bilirubin from the liver to the intestines. It is then eliminated in the feces.

Patients with Gilbert syndrome have around 30% of normal bilirubin-UGT enzyme performance, which results in a decreased rate of unconjugated bilirubin glucuronidation.

Gilbert syndrome is a phenotypic effect caused by several different genotypic variants of the gene for the enzyme that converts bilirubin to the conjugated form. It is usually characterized by increased blood bilirubin levels, but it can also be characterized by mild jaundice due to increased unconjugated bilirubin.

Gilbert's syndrome is characterized by a 70–80% decrease in the enzyme's glucuronidation activity *(UGT1A1).* The *UGT1A1* gene is found on chromosome 2 in humans. More than 100 polymorphisms in the *UGT1A1* gene, identified as *UGT1A1*n* (where n is the general chronological order of discovery), have been discovered, either in the gene or in the promoter region. A *TATA* box promoter region is related to *UGT1A1*; this area most typically comprises the genetic sequence *A (TA) 6TAA.* Across many groups, this variant accounts for nearly half of all alleles. However, there are several allelic polymorphic variants of this region, the most common of which results from adding another dinucleotide repeat *TA* to the promoter region, resulting in *A (TA)7TAA,* also known as *UGT1A1*28;* this common variant accounts for about 40% of alleles in some populations but is seen less frequently, around 3% of alleles, in Southeast and East Asian people and Pacific Islanders.

Gilbert syndrome is most typically associated with homozygous *A (TA) 7TAA* alleles in most populations. In 94% of GS cases, two more glucuronosyltransferase enzymes, *UGT1A6* (which is rendered 50% inactive) and *UGT1A7* (which is rendered 83% ineffective), are similarly impacted. Gilbert syndrome can, however, occur without *TATA* box promoter polymorphic variants; in some populations, particularly healthy Southeast and East Asians, heterozygote missense mutations in the actual gene coding region (such as Gly71Arg, also known as *UGT1A1*6*, Tyr486Asp, also known as *UGT1A1*7*, and Pro364Leu, also known as *UGT1A1*73*), are more frequently associated with significantly higher bilirubin levels Gilbert's syndrome is classified as a minor inborn error of metabolism because of its effects on drug and bilirubin breakdown, as well as its genetic inheritance.

CONCLUSION

Higher levels of unconjugated bilirubin in the blood without any additional signs of liver disease or red blood cell disintegration are used to make the diagnosis. It is then eliminated in the feces. Genetics Gilbert syndrome is a phenotypic effect caused by several different genotypic variants of the gene for the enzyme that converts bilirubin to the conjugated form. Gilbert's syndrome is characterized by a 70-80% decrease in the enzyme's

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