## DISEASE: Gilbert syndrome (NON RARE IN EUROPE)

ish substance produced during the normal breakdown of old red blood cells. Bilirubin travels through the bloodstream to the liver where it is processed and removed from the blood. The symptom of cholemia or hyperbilirubinemia (high bilirubin levels) is jaundice or yellowing of the skin and whites of the eyes. It is generally a mild condition and does not usually require treatment. Times of jaundice are sometimes induc in children and adults during infections and illness, lack of sleep, fasting (stretches of not eating), menstruation cycles, and stress and may be accompanied by mild abdominal pain or nausea. The jaundic during these times goes away on its own after the person recovers. In the newborn period, jaundice can be	NAME:	Gilbert syndrome (NON RARE IN EUROPE)
DESCRIPTION: unless a person or newborn has a severe bout of jaundice and the yellow in the skin or eyes is noted. Treatment is only necessary if bilirubin levels are very high.   More males than females have been diagnosed with Gilbert syndrome. This condition is caused by mutations in the UGT1A1 gene. Gilbert syndrome is inherited or passed through families in an autosomal recessive manner. This means that to be affected, an individual must have two copies of the changed gene that causes the condition. An affected person has then inherited one changed gene from each of his or here.	DESCRIPTION:	bloodstream to the liver where it is processed and removed from the blood. The symptom of cholemia or hyperbilirubinemia (high bilirubin levels) is jaundice or yellowing of the skin and whites of the eyes. It is generally a mild condition and does not usually require treatment. Times of jaundice are sometimes induced in children and adults during infections and illness, lack of sleep, fasting (stretches of not eating), menstruation cycles, and stress and may be accompanied by mild abdominal pain or nausea. The jaundice during these times goes away on its own after the person recovers. In the newborn period, jaundice can be longer-lasting and more severe. Diagnosis often occurs by accident when blood is drawn for another reason unless a person or newborn has a severe bout of jaundice and the yellow in the skin or eyes is noted. Treatment is only necessary if bilirubin levels are very high. More males than females have been diagnosed with Gilbert syndrome. This condition is caused by mutations in the UGT1A1 gene. Gilbert syndrome is inherited or passed through families in an autosomal recessive manner. This means that to be affected, an individual must have two copies of the changed gene that causes the condition. An affected person has then inherited one changed gene from each of his or her parents. Each child of two carrier parents has a 25% chance of inheriting both changed genes and therefore the condition. A genetic counselor can provide an understanding of the underlying cause as well as

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- Gilbert syndrome / Irinotecan sensitivity (homozygous A(TA)7TAA allele) Pharmacogenetics
- Gilbert syndrome / Irinotecan sensitivity (homozygous A(TA)7TAA allele)

## **Related Laboratories**

- Centre de Génétique Humaine CHU Sart-Tilman
- Centre de Génétique Humaine Erasme ULB
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
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