A CASE OF GILBERT’S SYNDROME

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Abstract
A middle aged lady presented with recurrent jaundice with normal SGPT, serum alkaline phosphatase, serum albumin and prothrombin time. Haemolysis was excluded by normal haemoglobin, peripheral blood film and reticulocyte count and finally she was diagnosed to have Gilbert’s syndrome.

Key words: Gilbert’s Syndrome, Hyperbilirubinaemia.


Discussion:
Gilbert’s syndrome is named after Augustin Gilbert (1858-1927), a French physician 1. It is defined as benign, familial, mild, unconjugated hyperbilirubinaemia not due to haemolysis and with normal routine tests of hepatic function and liver histology. It affects some 2-5 % population.2

Unconjugated bilirubin is lipid-soluble. Uridine diphosphateglucuronosyl transferase (UGT) is the enzyme that converts unconjugated bilirubin to conjugated bilirubin monoglucuronide and diglucuronide, makes it water soluble and allows its excretion into the bile. The gene expressing bilirubin UGT is on chromosome 2, having 5 exons and the promoter region (TATAA box) 3,4.

Patients with Gilbert’s syndrome have a deficiency in bilirubin UGT activity – about 30% of normal. It is inherited as autosomal recessive, that is, the patients are homozygous for this abnormality 5. The promoter region of the gene encoding bilirubin UGT – A(TA)6TAA has an additional TA dinucleotide – A(TA)7TAA 6,7. The condition may be diagnosed incidentally at a routine medical examination or when blood being examined for another reason, for instance, viral hepatitis. Jaundice is mild and...
intermittent. Bilirubin levels are most often <3 mg/dl. Deepening may follow an intercurrent infection or fasting and is associated with malaise, nausea and often discomfort over liver. There is no other abnormal physical sign.

Specialist diagnostic tests include the increase in serum bilirubin on fasting or following intravenous nicotinic acid which raises the osmotic fragility of RBC and the fall on taking phenobarbitone which induces hepatic conjugating enzymes.

However, Gilbert’s syndrome is usually diagnosed easily without recourse to these specialist methods. The demonstration of a raised bilirubin level that is predominantly unconjugated, with normal liver enzymes and no evidence of haemolysis, is usually sufficient.

Gilbert’s syndrome has an excellent prognosis. Patients have a normal life expectancy and reassurance is the only necessary treatment. Hyperbilirubinaemia is life long and not associated with increased morbidity. Serum bilirubin may be reduced by phenobarbitone but, as jaundice is rarely obvious, few patients will get cosmetic benefit from this treatment. Patients should be warned that jaundice can follow an intercurrent infection, repeated vomiting or missed meal.

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References: