

INVESTIGATION OF GILBERT'S SYNDROME

INTRODUCTION

Gilbert's syndrome is a common (occurring in ~5% of population) and benign condition characterized by recurrent episodes of mild jaundice, the total bilirubin concentration being less than 100 $\mu\text{mol/L}$. In Gilbert's syndrome other tests of liver function are normal and liver histology is also normal. It is important to recognize this syndrome because the hyperbilirubinaemia may be mistaken as a sign of liver disease.

Jaundice in Gilbert's syndrome is often associated with inter-current illness and caloric deprivation. The mechanism of jaundice is unclear but decreased bilirubin uptake by the liver is commonly present and decreased conjugation is demonstrable in some individuals.

In addition to showing that the hyperbilirubinaemia is due to unconjugated bilirubin and excluding haemolysis, caloric deprivation or fasting may be used sometimes to confirm a diagnosis of Gilbert's syndrome.

LABORATORY INVESTIGATION

Reports of liver function tests to GP's are examined for isolated hyperbilirubinaemia (total bilirubin >25 $\mu\text{mol/l}$) and conjugated bilirubin is added when the total bilirubin is ≥ 50 $\mu\text{mol/l}$.

A comment is added to the report providing:

- Clinical details given on request form do not suggest history of liver pathology
- The conjugated bilirubin fraction accounts for $<20\%$ of the total bilirubin level i.e. increased total bilirubin in Gilbert's syndrome should be due largely ($>80\%$) to unconjugated bilirubin.
- Any haematology investigations done at the same time do not suggest evidence of haemolysis (low haemoglobin level, increased reticulocyte count).

'If no liver pathology or intravascular haemolysis, raised unconjugated bilirubin consistent with Gilberts syndrome'

It is rarely necessary but if further confirmation of Gilbert's is required the response to fasting can be studied. Following a 24-hour fast (or 48 hours of a low caloric diet) there is a 2 to 3 fold increase in unconjugated bilirubin in subjects with Gilbert's syndrome. Much smaller increases, or no increase at all, occur in normal individuals.