

Evaluation of Isolated Hyperbilirubinaemia

(Bilirubin > 30 $\mu\text{mol/L}$; other LFTs normal)

Common causes

- Fasting (< 40 $\mu\text{mol/L}$)
- Gilbert's syndrome
- Haemolytic disease
- Dubin-Johnson syndrome

Fasting

A calorie intake of < 400 per 24 hours may be associated with a two- to three-fold rise in **unconjugated** bilirubin, but values > 35 $\mu\text{mol/L}$ are uncommon.

Gilbert's syndrome

Unconjugated hyperbilirubinaemia of 25–100 $\mu\text{mol/L}$. Occurs in 2–7% of the general population (the ratio of occurrence in males to females is 2–7:1). Characterised by intermittent mild jaundice with bilirubin values increasing during fasting and non-specific illness. Inherited as genetic disorder and is harmless in adults due mainly to a conjugation defect.

Haemolysis

Chronic haemolysis can result in an **unconjugated** hyperbilirubinaemia of 50–100 $\mu\text{mol/L}$; values in excess of 100 $\mu\text{mol/L}$ suggest a second process (e.g. Gilbert's syndrome). Usually associated with reticulocytosis, elevated LD, and low serum haptoglobin levels.

Dubin-Johnson syndrome

Rare autosomal recessive disorder with fluctuating **conjugated** hyperbilirubinaemia (up to 100 $\mu\text{mol/L}$). Due to inability to excrete conjugated bilirubin; harmless to patient.

Laboratory evaluation

Aetiology is usually obvious from clinical picture; if the cause is obscure, the following tests are helpful:

- Bilirubin fractionation
- Full blood picture
- Reticulocyte count
- Serum haptoglobin
- Serum LD
- Gilbert's syndrome genotyping (non-rebateable)