

Hereditary hyperbilirubinemias

All autosomal recessive.

1 Gilbert syndrome

Mildly ↓ UDP-glucuronosyltransferase conjugation and impaired bilirubin uptake. Asymptomatic or mild jaundice usually with stress, illness, or fasting. ↑ unconjugated bilirubin without overt hemolysis.

Relatively common, benign condition.

2 Crigler-Najjar syndrome, type I

Absent UDP-glucuronosyltransferase. Presents early in life, but some patients may not have neurologic signs until later in life. Findings: jaundice, kernicterus (bilirubin deposition in brain), ↑ unconjugated bilirubin. Treatment: plasmapheresis and phototherapy (does not conjugate UCB; but does ↑ polarity and ↑ water solubility to allow excretion). Liver transplant is curative.

Type II is less severe and responds to phenobarbital, which ↑ liver enzyme synthesis.

3 Dubin-Johnson syndrome

Conjugated hyperbilirubinemia due to defective liver excretion. Grossly black (Dark) liver. Benign.

4 Rotor syndrome is similar, but milder in presentation without black (Regular) liver. Due to impaired hepatic uptake and excretion.

