Fat digestion is a tricky process, compared to carbohydrate or protein digestion. Nevertheless, fats are a valuable component of the human diet, and we have evolved an efficient system for extracting the fat from our food. Because fats are not water-soluble, they are first emulsified by the secretion of bile acids from the gallbladder, before they are broken down by digestive enzymes.

Bile acids resemble steroids in structure, and are derived from cholesterol. They act as powerful detergents, and therefore are highly toxic to surrounding tissues. Because of this, their production and secretion is tightly controlled in the digestive tract.

Cholestatic syndrome is a general term encompassing a range of disorders in which bile excretion from the liver is blocked. One of the most well-known of these is newborn cholestasis, or “jaundice”. Cholestatic syndromes afflict both adults and children alike, with varying disease severity and symptoms.

What causes the disruption of bile excretion in the liver? In his review on “Familial cholestatic syndromes”, Richard Thompson examines this class of disorders from a molecular perspective. Focusing on the inherited syndromes, Thompson points out the genetic hot spots suspected to be linked to the such diverse diseases as Alagille Syndrome, ARC Syndrome and BSEP deficiency.

“In the last 10 years, our understanding of the molecular mechanisms involved in bile formation and cholestasis has improved dramatically,” states Thompson. Despite the complexity of these disorders, the identity of the genetic culprits is becoming clearer. This improved knowledge will speed up diagnosis and provides clues for new treatment design.

References