ABSTRACT

Cholesterol 7α-hydroxylase (CYP7A1) is an enzyme catalyzing the first step of conversion of cholesterol to bile acids. The enzyme activity is regulated to supply enough bile salts necessary for absorption of fats in the intestine. In some species it contributes to cholesterol elimination from the body when dietary cholesterol intake is high and, in such a way, protects against the development of hypercholesterolemia. CYP7A1 activity can be therapeutically affected by administration of bile acid sequestrants that increase the enzyme activity and thus lower cholesterolemia, and also by administration of bile salts. The enzyme deficiency in humans results in hypercholesterolemia. Several single nucleotide polymorphisms were identified in gene encoding CYP7A1 in humans. They form three large haplotype blocks. The most attention has been paid to the -203A>C polymorphism that has an impact on cholesterol and lipoprotein concentrations, on the response of cholesterolemia to dietary intervention and on the response to hypolipidemic drugs.

Key words:

Bile acids, cholesterol, cholesterol 7α-hydroxylase, diet, genetics, treatment of hypercholesterolemia