



Need to talk? Call us **0845 2412173** Monday to Friday  
9am to 5pm



Supporting those affected by  
Inherited Metabolic Disorders

# Cholesterol 7alpha-Hydroxylase Deficiency

CYP7A1 Deficiency is a rare disorder that affects bile acid synthesis. It is caused by a defect in a gene called CYP7A1 which is essential to produce an enzyme known as cholesterol 7alpha-hydroxylase. This enzyme catalyses the first step in the breakdown (catabolism) of cholesterol and the creation of bile acids. Bile acids are found in the liver and are formed from cholesterol. As well as performing other roles, they promote the flow and excretion of bile and help the intestine to absorb fat and fat-soluble vitamins. A problem in forming bile acids causes abnormal bile acids, cholesterol, and other substances to accumulate in the body causing damage to certain organs. Symptoms include gallstones, heart and peripheral vascular disease, Unlike other bile acid disorders, CYP7A1 Deficiency does not usually cause liver disease.

## Synonyms

Alternative names for this condition are:

- CYP7A1 Deficiency

Further information about this condition is available from Climb.

## Disclaimer

[Please read our disclaimer and information on data protection.](#)



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