CYP27A1 Gene

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Cytochrome P450 Family 27 Subfamily A Member 1

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1. Normal Function

The *CYP27A1* gene is a member of the cytochrome P450 gene family. Enzymes produced from the cytochrome P450 genes are involved in the formation and breakdown of various molecules and chemicals within cells. The *CYP27A1* gene provides instructions for producing an enzyme called sterol 27-hydroxylase. This enzyme is located in the energy-producing centers of cells (mitochondria), where it is involved in the pathway that breaks down cholesterol to form acids used to digest fats (bile acids). Specifically, sterol 27-hydroxylase breaks down cholesterol to form a bile acid called chenodeoxycholic acid. The formation of bile acids from cholesterol is the body's main pathway for cholesterol removal. Sterol 27-hydroxylase plays a key role in maintaining normal cholesterol levels in the body.

2. Health Conditions Related to Genetic Changes

2.1 Cerebrotendinous Xanthomatosis

At least 90 mutations that cause cerebrotendinous xanthomatosis have been identified in the *CYP27A1* gene. Cerebrotendinous xanthomatosis is a disorder characterized by abnormal storage of fats (lipids) in many areas of the body. Most *CYP27A1* gene mutations change one protein building block (amino acid) in the sterol 27-hydroxylase enzyme. The most common mutation changes the amino acid arginine to the amino acid cysteine at position 362 in the protein (written as Arg362Cys or R362C). Changes in amino acids typically disrupt the normal function of the protein and impair its ability to help form chenodeoxycholic acid. Other mutations cause no functional enzyme to be made. As a result, other molecules are formed by an alternative pathway. A molecule called cholestanol, which is similar to cholesterol, is produced and accumulates in blood and tissues. Cholesterol also accumulates in tissues, but levels in blood are typically normal. The accumulation of cholesterol and cholestanol throughout the body's tissues causes the signs and symptoms of cerebrotendinous xanthomatosis.

3. Other Names for This Gene

- 5-beta-cholestane-3-alpha, 7-alpha, 12-alpha-triol 27-hydroxylase
- CP27
- CP27A_HUMAN
- CTX
- CYP27
- cytochrome P-450C27/25
- cytochrome P450, family 27, subfamily A, polypeptide 1
- cytochrome P450, subfamily XXVIIA (steroid 27-hydroxylase, cerebrotendinous xanthomatosis), polypeptide 1
- sterol 27-hydroxylase
- vitamin D(3) 25-hydroxylase

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