ETHE1 Gene

Subjects: Genetics & Heredity

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ETHE1, persulfide dioxygenase

genes

1. Normal Function

The *ETHE1* gene provides instructions for making an enzyme that is active in mitochondria, which are the energy-producing centers in cells. The ETHE1 enzyme is part of a pathway that breaks down a molecule called sulfide (H_2S) in mitochondria. Sulfide is produced in the body's tissues as part of normal cell processes, and it is also released by bacteria living in the gastrointestinal system (gut).

At low levels, sulfide is critical for normal cell functioning. However, this molecule becomes toxic at high levels, interfering with numerous cell activities. For example, excess sulfide interferes with mitochondrial energy production by blocking (inhibiting) an enzyme complex called cytochrome C oxidase (COX). This complex normally carries out one of the final steps in the process of energy production in mitochondria.

2. Health Conditions Related to Genetic Changes

2.1 Ethylmalonic Encephalopathy

More than 30 mutations in the *ETHE1* gene have been identified in people with ethylmalonic encephalopathy. This rare condition affects many parts of the body, including the nervous system, blood vessels, and intestines. Signs and symptoms include delayed development, abnormal movements, rashes of tiny red spots under the skin (petechiae), blue discoloration of the hands and feet (acrocyanosis), and chronic diarrhea.

Most of the mutations that cause ethylmalonic encephalopathy lead to the production of nonfunctional versions of the ETHE1 enzyme or prevent cells from making any of this enzyme. A shortage of functional enzyme prevents sulfide from being broken down normally, allowing this molecule to accumulate in cells. The buildup of sulfide inhibits the activity of COX, which disrupts mitochondrial energy production and damages tissues and organs throughout the body. Researchers believe that the effects of excess sulfide in the brain, muscles, blood vessels, and lining of the intestines underlie most of the major features of ethylmalonic encephalopathy.

2.2 Leigh Syndrome

3. Other Names for This Gene

- ETHE1_HUMAN
- ethylmalonic encephalopathy 1
- Ethylmalonic encephalopathy protein 1
- hepatoma subtracted clone one
- HSCO
- YF13H12

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