MT-TE Gene

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mitochondrially encoded tRNA glutamic acid

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1. Introduction

The *MT-TE* gene provides instructions for making a molecule called a transfer RNA (tRNA), which is a chemical cousin of DNA. Transfer RNAs help assemble protein building blocks (amino acids) into functioning proteins. The *MT-TE* gene provides instructions for making a specific form of tRNA that is designated as $tRNA^{Glu}$. During protein assembly, this molecule attaches to the amino acid glutamic acid (Glu) and inserts it into the appropriate locations in the growing protein.

The $tRNA^{Glu}$ molecule is present only in cellular compartments called mitochondria. These structures convert energy from food into a form that cells can use. Through a process called oxidative phosphorylation, mitochondria use oxygen, simple sugars, and fatty acids to create adenosine triphosphate (ATP), the cell's main energy source. The $tRNA^{Glu}$ molecule is involved in the assembly of proteins that carry out oxidative phosphorylation.

In certain cells in the pancreas, called beta cells, mitochondria also play a role in controlling the amount of sugar (glucose) in the bloodstream. In response to high glucose levels, mitochondria help trigger the release of a hormone called insulin. Insulin regulates blood sugar levels by controlling how much glucose is passed from the blood into cells to be converted into energy.

2. Health Conditions Related to Genetic Changes

2.1. Maternally inherited diabetes and deafness

A mutation in the *MT-TE* gene has been found in a small number of people with maternally inherited diabetes and deafness (MIDD). People with this condition have diabetes and sometimes hearing loss, particularly of high tones. Affected individuals may also have muscle weakness (myopathy) and problems with their eyes, heart, or kidneys. The mutation involved in this condition replaces the DNA building block (nucleotide) thymine with the nucleotide cytosine at position 14709 (written as T14709C). This mutation likely impairs the ability of mitochondria to help trigger insulin release. In affected individuals, diabetes results when the beta cells do not produce enough insulin to regulate blood sugar effectively. Researchers have not determined how the T14709C mutation leads to hearing loss or the other features of MIDD.

2.2. Other disorders

Mutations in the *MT-TE* gene are also involved in infantile transient mitochondrial myopathy (also known as benign COX deficiency myopathy). This rare condition occurs within the first few months of life and causes severe muscle weakness, poor muscle tone (hypotonia), and buildup of a chemical called lactic acid in the body (lactic acidosis). Affected infants often have difficulty feeding and need support from a machine to help them breathe. The signs and symptoms improve after several months, and most affected individuals show no symptoms of the condition by age 2 or 3.

The mutations involved in infantile transient mitochondrial myopathy change single nucleotides in mitochondrial DNA. Specifically, the nucleotide thymine at position 14674 is replaced by the nucleotide cytosine or guanine (written as T14674C or T14674G, respectively). These mutations impair oxidative phosphorylation. As a result, muscle cells cannot produce enough energy, leading to the muscle problems that affect infants with infantile transient mitochondrial myopathy. It is unknown why only muscles are involved or how affected infants recover from the condition.

3. Other Names for This Gene

- MTTE
- trnE

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