

# MT-TV Gene

Subjects: **Genetics & Heredity**

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mitochondrially encoded tRNA valine

genes

## 1. Introduction

The *MT-TV* gene provides instructions for making a particular type of RNA, a molecule that is a chemical cousin of DNA. This type of RNA, called transfer RNA (tRNA), helps assemble protein building blocks known as amino acids into full-length, functioning proteins. The *MT-TV* gene provides instructions for a specific form of transfer RNA that is designated as tRNA<sup>Val</sup>. This molecule attaches to a particular amino acid, valine (Val), and inserts it into the appropriate locations in many different proteins.

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

## 2. Health Conditions Related to Genetic Changes

### 2.1. Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes

Mutations in the *MT-TV* gene are a very rare cause of mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS). Most cases of MELAS are caused by mutations in other mitochondrial genes, but a small number of cases resulting from mutations in the *MT-TV* gene have been reported. At least two mutations have been identified, each of which alters a single DNA building block (nucleotide) in the gene. One of these mutations replaces the nucleotide guanine with the nucleotide adenine at gene position 1642 (written as G1642A). Another mutation changes the nucleotide guanine to the nucleotide adenine at position 1644 (G1644A). Changes in the *MT-TV* gene may reduce the amount of tRNA<sup>Val</sup> available to assemble proteins within mitochondria. Because these proteins are needed for oxidative phosphorylation, *MT-TV* mutations may impair the ability of mitochondria to produce energy. Researchers have not determined how changes in mitochondrial energy production can lead to the specific features of MELAS.

### 2.2. Other disorders

A few other mutations in the *MT-TV* gene have been reported, most of which change single nucleotides in the gene. These mutations are responsible for a variety of signs and symptoms chiefly affecting the muscles and nervous system. Medical problems associated with *MT-TV* mutations have included recurrent migraine headaches, muscle weakness and problems with movement, poor coordination, seizures, hearing loss, learning disabilities, and loss of intellectual function (dementia). A heart condition called cardiomyopathy, which weakens and enlarges the heart muscle, also has been reported in a small number of affected individuals.

Changes in the *MT-TV* gene have been identified as a rare cause of Leigh syndrome, a progressive brain disorder that typically appears in infancy or early childhood. In a few cases, *MT-TV* mutations were found in people with signs and symptoms that did not appear until adulthood. Affected individuals may experience vomiting, seizures, delayed development, muscle weakness, and problems with movement. Heart disease, kidney problems, and difficulty breathing can also occur in people with this disorder.

It is unclear why changes in the *MT-TV* gene can cause such a large variety of signs and symptoms. Even within a single family, affected individuals may have different health problems caused by the same genetic change.

## 3. Other Names for This Gene

- MTTV
- tRNA valine
- tRNA-Val, mitochondrial

## References

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