TRNT1 Gene

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tRNA nucleotidyl transferase 1: The TRNT1 gene provides instructions for making a protein involved in the production (synthesis) of other proteins.

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1. Normal Function

The *TRNT1* gene provides instructions for making a protein involved in the production (synthesis) of other proteins. During protein synthesis, a molecule called transfer RNA (tRNA) helps assemble protein building blocks (amino acids) into a chain that forms the protein. Each tRNA carries a specific amino acid to the growing chain. The TRNT1 protein modifies tRNAs by adding a series of three DNA building blocks (nucleotides), called a CCA trinucleotide, to the molecule. This modification is essential for the correct amino acid to be attached to each tRNA.

While most protein synthesis occurs in the fluid surrounding the nucleus (cytoplasm), some proteins are synthesized in cell structures called mitochondria, which are the energy-producing centers in cells. Many mitochondrial proteins form groups (complexes) that carry out the reactions that produce energy. Separate tRNA molecules are used to build proteins in the cytoplasm and mitochondria. The TRNT1 protein attaches the CCA trinucleotide to both cytoplasmic and mitochondrial tRNA molecules.

2. Health Conditions Related to Genetic Changes

2.1. TRNT1 deficiency

More than 20 *TRNT1* gene mutations have been found to cause TRNT1 deficiency, a condition with a range of signs and symptoms that affect many body systems. Features can include a blood disorder called sideroblastic anemia, recurrent fevers, a shortage of immune cells called B cells that leads to impairment of the immune system (immunodeficiency), delayed development of speech and motor skills, and eye abnormalities that cause vision problems. The severity of the condition varies among affected individuals.

The *TRNT1* gene mutations that cause TRNT1 deficiency lead to a shortage (deficiency) of functional TRNT1 protein. As a result, the addition of the CCA trinucleotide to tRNA molecules is impaired. Researchers suspect that without the modification, tRNAs are less able to participate in protein synthesis. Studies show that deficiency of TRNT1 prevents the formation of certain mitochondrial protein complexes that are involved in energy production. It is unclear if cytoplasmic protein synthesis is also affected. The reduction of energy could damage cells in many body systems, leading to the varied signs and symptoms of TRNT1 deficiency. Researchers believe that mutations that cause a greater impairment of TRNT1 function lead to more severe signs and symptoms.

3. Other Names for This Gene

- ATP(CTP):tRNA nucleotidyltransferase
- CCA tRNA nucleotidyltransferase 1, mitochondrial isoform 1
- CCA tRNA nucleotidyltransferase 1, mitochondrial isoform 2
- CCA-adding enzyme
- CCA1
- CGI-47
- mitochondrial CCA-adding tRNA-nucleotidyltransferase
- mt CCA-adding enzyme
- mt tRNA adenylyltransferase

- mt tRNA CCA-diphosphorylase
- mt tRNA CCA-pyrophosphorylase
- MtCCA
- RPEM
- SIFD
- tRNA CCA nucleotidyl transferase 1
- tRNA nucleotidyl transferase, CCA-adding, 1
- tRNA-nucleotidyltransferase 1, mitochondrial

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