TSEN34 Gene

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tRNA splicing endonuclease subunit 34

genes

1. Normal Function

The *TSEN34* gene provides instructions for making one part (subunit) of an enzyme called the tRNA splicing endonuclease complex. This complex helps process several types of RNA molecules, which are chemical cousins of DNA.

The tRNA splicing endonuclease complex is particularly important for the normal processing of a form of RNA known as transfer RNA (tRNA). tRNA molecules help assemble protein building blocks called amino acids into full-length proteins. However, before they can assemble proteins, tRNAs must be processed into mature molecules. In particular, regions called introns need to be removed from some tRNAs for the molecules to be functional. The tRNA splicing endonuclease complex recognizes and then removes introns to help produce mature tRNA molecules.

Studies suggest that the tRNA splicing endonuclease complex may also be involved in processing another form of RNA known as messenger RNA (mRNA). mRNA serves as a genetic blueprint for making proteins. Researchers suspect that the tRNA splicing endonuclease complex cuts (cleaves) one end of mRNA molecules so a string of adenines (one of the building blocks of RNA) can be added. This process is known as polyadenylation, and the string of adenines is known as a poly(A) tail. The poly(A) tail signals the stopping point for protein production and protects mRNA from being broken down before protein production occurs.

2. Health Conditions Related to Genetic Changes

2.1. Pontocerebellar Hypoplasia

At least one mutation in the *TSEN34* gene has been identified in people with a disorder of brain development called pontocerebellar hypoplasia. The major features of this condition include delayed development, problems with movement, and intellectual disability. *TSEN34* gene mutations cause a small percentage of all cases of a form of the disorder designated pontocerebellar hypoplasia type 2 (PCH2). When PCH2 results from *TSEN34* gene mutations, it is sometimes categorized more specifically as PCH2C.

The mutation that causes PCH2C replaces the amino acid arginine with the amino acid tryptophan at position 58 in the TSEN34 protein (written as Arg58Trp or R58W). This mutation impairs the function of the tRNA splicing endonuclease complex, which likely disrupts the processing of RNA molecules and affects the production of many types of proteins. Before birth, these changes appear to have the most severe impact on fast-growing tissues, such as those in the brain. However, it is unknown exactly how reduced function of the tRNA splicing endonuclease complex leads to abnormal brain development in people with this condition.

3. Other Names for This Gene

- LENG5
- SEN34
- SEN34_HUMAN
- SEN34L
- tRNA splicing endonuclease 34
- tRNA splicing endonuclease 34 homolog (S. cerevisiae)
- TSEN34 tRNA splicing endonuclease subunit

References

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