TREX1 Gene

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Contributor: Rui Liu

Three prime repair exonuclease 1: The TREX1 gene provides instructions for making the 3-prime repair exonuclease 1 enzyme.

genes

1. Normal Function

The *TREX1* gene provides instructions for making the 3-prime repair exonuclease 1 enzyme. This enzyme is a DNA exonuclease, which means that it trims molecules of DNA by removing DNA building blocks (nucleotides) from the ends of the molecules. In this way, it breaks down unneeded DNA molecules or fragments that may be generated during copying (replication) of cells' genetic material in preparation for cell division. These fragments may also be generated during DNA repair, cell death (apoptosis), and other processes.

2. Health Conditions Related to Genetic Changes

2.1. Aicardi-Goutières syndrome

At least 82 mutations in the *TREX1* gene have been identified in people with Aicardi-Goutières syndrome, a disorder that involves severe brain dysfunction (encephalopathy), skin lesions, and other health problems. Most of these mutations are believed to prevent the production of the 3-prime repair exonuclease 1 enzyme. Researchers suggest that the absence of this enzyme results in an accumulation of unneeded DNA and RNA in cells. These DNA and RNA molecules may be mistaken by cells for the genetic material of viral invaders, triggering immune system reactions that damage the brain, skin, and other organs and systems and result in the signs and symptoms of Aicardi-Goutières syndrome.

2.2. Other disorders

Mutations in the *TREX1* gene have also been identified in people with other disorders involving the immune system. These disorders include a chronic inflammatory disease called systemic lupus erythematosus (SLE), including a rare form of SLE called chilblain lupus that mainly affects the skin. Features of SLE, especially chilblain lupus, often also occur in people with Aicardi-Goutières syndrome (described above).

TREX1 gene mutations have also been found in people with a disorder called autosomal dominant retinal vasculopathy with cerebral leukodystrophy, which affects the brain and the blood vessels in the specialized light-

sensitive tissue that lines the back of the eye (the retina).

As in Aicardi-Goutières syndrome, absence or impaired function of the 3-prime repair exonuclease 1 enzyme may cause immune system dysfunction that damages the brain, skin, blood vessels, and other parts of the body. It is not clear why mutations in the same gene cause several different disorders.

Systemic lupus erythematosus

3. Other Names for This Gene

- 3' repair exonuclease 1
- 3'-5' exonuclease TREX1
- AGS1
- CRV
- deoxyribonuclease III, dnaQ/mutD-like
- DKFZp434J0310
- DNase III
- DRN3
- HERNS
- three prime repair exonuclease 1 isoform a
- three prime repair exonuclease 1 isoform b
- TREX1_HUMAN

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