ADA Gene

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Adenosine deaminase

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1. Normal Function

The *ADA* gene provides instructions for producing the enzyme adenosine deaminase. This enzyme is produced in all cells, but the highest levels of adenosine deaminase occur in immune system cells called lymphocytes, which develop in lymphoid tissues. These lymphoid tissues include the thymus, which is a gland located behind the breastbone, and lymph nodes, which are found throughout the body. Lymphocytes form the immune system, which defends the body against potentially harmful invaders, such as viruses or bacteria.

The function of the adenosine deaminase enzyme is to eliminate a molecule called deoxyadenosine, which is generated when DNA is broken down. Adenosine deaminase converts deoxyadenosine, which is toxic to lymphocytes, to another molecule called deoxyinosine, which is not harmful.

2. Health Conditions Related to Genetic Changes

2.1. Adenosine deaminase deficiency

More than 70 mutations in the *ADA* gene have been identified. Most of these mutations result in the substitution of one protein building block (amino acid) for another amino acid in the adenosine deaminase enzyme. Other mutations cause the enzyme to be unstable or prevent it from being produced at all.

These mutations result in the absence or deficiency of the adenosine deaminase enzyme in cells, preventing the normal breakdown of deoxyadenosine. A buildup of this toxic compound interferes with the development and maintenance of lymphocytes, resulting in severe combined immunodeficiency (SCID), which is characteristic of adenosine deaminase deficiency.

3. Other Names for This Gene

- ADA_HUMAN
- adenosine aminohydrolase

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