CAT Gene

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catalase

genes

1. Normal Function

The *CAT* gene provides instructions for making pieces (subunits) of an enzyme called catalase. Four identical subunits, each attached (bound) to an iron-containing molecule called a heme group, form the functional enzyme.

Catalase is active in cells and tissues throughout the body, where it breaks down hydrogen peroxide (H_2O_2) molecules into oxygen (O_2) and water (H_2O). Hydrogen peroxide is produced through chemical reactions within cells. At low levels, it is involved in several chemical signaling pathways, but at high levels it is toxic to cells. If hydrogen peroxide is not broken down by catalase, additional reactions convert it into compounds called reactive oxygen species that can damage DNA, proteins, and cell membranes.

2. Health Conditions Related to Genetic Changes

2.1. Acatalasemia

At least 13 mutations in the *CAT* gene have been found to cause acatalasemia, a condition characterized by very low catalase activity. Many people with acatalasemia never have any related health problems, although the condition has occasionally been associated with open sores (ulcers) inside the mouth leading to the death of soft tissue (gangrene). Acatalasemia also appears to increase the risk of developing type 2 diabetes (the most common form of diabetes) and is a potential risk factor for other common, complex diseases.

The mutations that cause acatalasemia occur in both copies of the *CAT* gene in each cell, and they reduce the activity of catalase to less than 10 percent of normal. A shortage of this enzyme can allow hydrogen peroxide to build up to toxic levels in certain cells. For example, hydrogen peroxide produced by bacteria in the mouth may accumulate in and damage soft tissues, leading to mouth ulcers and gangrene. A buildup of hydrogen peroxide may also damage beta cells of the pancreas, which release a hormone called insulin that helps control blood sugar. Malfunctioning beta cells are thought to underlie the increased risk of type 2 diabetes in people with acatalasemia. It is unclear why some people have no health problems associated with a shortage of catalase activity.

A related condition called hypocatalasemia occurs when only one of the two copies of the CAT gene in each cell has a mutation. This single mutation reduces the activity of catalase by approximately half. Like acatalasemia, hypocatalasemia usually does not cause any health problems.

2.2. Other Disorders

Common variations (polymorphisms) in the *CAT* gene and in regions of DNA that regulate the gene's activity may be associated with the risk of developing certain common, complex diseases. For example, researchers are studying these polymorphisms as potential risk factors for type 2 diabetes and other disorders of blood sugar regulation. *CAT* gene polymorphisms may also be associated with high blood pressure (hypertension), a skin condition called vitiligo, thinning of the bones (osteoporosis), and elevated levels of cholesterol and other fats (lipids) in the blood, which increase the risk of heart attack and stroke. However, it is unclear how polymorphisms in the *CAT* gene impact catalase activity, and how changes in the activity of this enzyme might influence a person's risk of developing these diseases. A large number of genetic and lifestyle factors, many of which remain unknown, likely determine the risk of developing most common, complex conditions.

3. Other Names for This Gene

- CATA_HUMAN
- EC 1.11.1.6

References

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