SI gene

sucrase-isomaltase

Normal Function

The *SI* gene provides instructions for producing the enzyme sucrase-isomaltase. This enzyme is made in the cells that line the small intestine, where it is involved in breaking down the sugars sucrose (a sugar found in fruits, and also known as table sugar) and maltose (the sugar found in grains). This enzyme is also important for digesting starches, which are first broken down into sucrose and maltose. Sucrose and maltose are called disaccharides because they are each made up of two simple sugar molecules. Disaccharides must be broken down into simple sugar molecules to be used by the body.

The sucrase-isomaltase enzyme is found on the surface of the intestinal epithelial cells, which are cells that line the walls of the intestine. These cells have fingerlike projections called microvilli that absorb nutrients from food as it passes through the intestine. Based on their appearance, groups of these microvilli are known collectively as the brush border. The role of the sucrase-isomaltase enzyme is to break down sucrose and maltose into simple sugars so that they can be absorbed by microvilli into intestinal epithelial cells.

Health Conditions Related to Genetic Changes

Congenital sucrase-isomaltase deficiency

Many variants (also known as mutations) in the *SI* gene have been found to cause congenital sucrase-isomaltase deficiency. These variants disrupt the folding and processing of the sucrase-isomaltase enzyme, transportation of the enzyme within the intestinal epithelial cells, localization of the enzyme at the correct cell surface, or its normal functioning. An impairment in any of these cell processes results in a sucrase-isomaltase enzyme that cannot effectively break down sucrose, maltose, or other sugars (carbohydrates) from starchy food. Rather than being absorbed by the small intestine, the undigested sugars move to the large intestine (colon). Here, they attract water and are consumed by normal bacteria in the colon, causing intestinal discomfort in people with congenital sucrase-isomaltase deficiency.

Other Names for This Gene

- MGC131621
- MGC131622
- sucrase-isomaltase (alpha-glucosidase)
- SUIS_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SI (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6476[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SI%5BTIAB%5D%29+OR+%28sucrase-isomaltase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SUCRASE-ISOMALTASE; SI (https://omim.org/entry/609845)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6476)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SI[gene])

References

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and polarized sorting act in concert to define a novel pathomechanismof congenital sucrase-isomaltase deficiency. J Biol Chem. 2006 May19;281(20):14393-9. doi: 10.1074/jbc.M513631200. Epub 2006 Mar 16. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16543230)

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- Sander P, Alfalah M, Keiser M, Korponay-Szabo I, Kovacs JB, Leeb T, Naim HY. Novel mutations in the human sucrase-isomaltase gene (SI) that cause congenitalcarbohydrate malabsorption. Hum Mutat. 2006 Jan;27(1):119. doi:10.1002/humu.9392. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16329100)

Genomic Location

The *SI* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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