Congenital sucrase-isomaltase deficiency

Description

Congenital sucrase-isomaltase deficiency is a rare genetic disorder that affects an individual's ability to digest certain sugars. People with this condition cannot break down the sugars sucrose and maltose. Sucrose (a sugar found in fruits, and also known as table sugar) and maltose (the sugar found in grains) are called disaccharides because they are made of two simple sugars. Disaccharides are broken down into simple sugars during digestion. Sucrose is broken down into glucose and another simple sugar called fructose, and maltose is broken down into two glucose molecules. People with congenital sucrase-isomaltase deficiency cannot break down the sugars sucrose and maltose, and other compounds made from these sugar molecules (carbohydrates).

Congenital sucrase-isomaltase deficiency usually becomes apparent after an infant is weaned and starts to consume fruits, juices, grains, and other starchy food. After ingestion of sucrose or maltose, an affected individual will typically experience stomach cramps, bloating, excess gas production, and diarrhea. These digestive problems can lead to failure to gain weight and grow at the expected rate (failure to thrive) and malnutrition.

Frequency

The prevalence of congenital sucrase-isomaltase deficiency is estimated to be 1 in 5,000 people of European descent. This condition is much more prevalent in the native populations of Greenland, Alaska, and Canada, where as many as 1 in 20 people may be affected.

Causes

Variants (also known as mutations) in the SI gene cause congenital sucrase-isomaltase deficiency. The SI gene provides instructions for producing the enzyme sucrase-isomaltase. This enzyme is found in the small intestine and is responsible for breaking down sucrose and maltose into their simple sugar components. These simple sugars are then absorbed by the small intestine. Variants that cause this condition alter the structure, disrupt the production, or impair the function of sucrase-isomaltase. These changes prevent the enzyme from breaking down sucrose and maltose. 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 the intestinal discomfort seen in individuals with congenital sucrase-isomaltase deficiency.

Learn more about the gene associated with Congenital sucrase-isomaltase deficiency

- SI

Inheritance

Congenital sucrase-isomaltase deficiency is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have variants. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they may not show signs and symptoms of the condition.

Other Names for This Condition

- Congenital sucrose intolerance
- Congenital sucrose-isomaltose malabsorption
- CSID
- Disaccharide intolerance I
- SI deficiency
- Sucrease-isomaltase deficiency

Additional Information & Resources

Genetic Testing Information


Genetic and Rare Diseases Information Center


Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials
• ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Congenital sucrase-isomaltase deficiency%22)

Catalog of Genes and Diseases from OMIM

• SUCRASE-ISOMALTASE DEFICIENCY, CONGENITAL; CSID (https://omim.org/entry/222900)

Scientific Articles on PubMed

• PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28congenital+sucrase-isomaltase+deficiency%5BTIAB%5D%29+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References


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