



Congenital Sucrase-Isomaltase Deficiency (CSID)

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Congenital sucrase-isomaltase deficiency (CSID) is a genetic disorder that affects a person's ability to digest certain sugars. People with this condition cannot break down the sugars sucrose and maltose, and other compounds made from simple sugar molecules (carbohydrates).

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.

Other names for CSID include genetic sucrase-isomaltase deficiency (GSID), congenital sucrose intolerance, congenital sucrose-isomaltase malabsorption, disaccharide intolerance I, SI deficiency, or sucrase-isomaltase deficiency.

Symptoms of CSID

Congenital sucrase-isomaltase deficiency usually becomes apparent in infants after they start to consume fruits, juices, and grains, and often diagnosed under the age of 18 months. After ingestion of sucrose or maltose, an affected person will typically experience watery diarrhea, bloating, excess gas production, abdominal pain ("stomach ache"), and malabsorption of other nutrients. Other symptoms may include nausea, vomiting, or reflux-like symptoms. In some children, these digestive problems can lead to failure to gain weight and grow at the expected rate (failure to thrive) and malnutrition. Most affected children are better able to tolerate sucrose and maltose as they get older.

Symptoms can vary for a variety of reasons including the timing of introduction of sucrose into a person's diet, and the amount of sugar and starch consumed. Infants who are breast-fed or fed lactose-containing formula will often not have symptoms of CSID until they ingest juices, solid foods, or medications that are sweetened by sucrose.

In some people symptoms may be milder than typically expected. Those with milder symptoms may not be diagnosed until later in childhood or in adulthood. In others with CSID the symptoms may mistakenly be thought due to something else, such as a functional gastrointestinal disorder like irritable bowel syndrome with diarrhea (IBS-D) or dyspepsia.

How common is CSID

The prevalence of CSID is still unknown and a subject of study and debate. Estimates of prevalence in people of European descent generally range from 1 in 500 to 1 in 2,000, and fewer African Americans are thought to be affected. The

condition is much more prevalent in the indigenous populations of Greenland, Alaska, and Canada, where as many as 1 in 10 to 1 in 30 people may be affected.

Nevertheless, more recent studies suggest that CSID may be more common than currently estimated. It is possible that some people remain undiagnosed and that the incidence is higher.

Causes of CSID

Mutations in a gene (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 involved in the digestion of sugar and starch. It is responsible for breaking down sucrose and maltose into their simple sugar components. These simple sugars are then absorbed by the small intestine.

In addition to genetic variations, other factors including dietary, gut motility, and nutritional interactions can affect the severity of symptoms.

Diagnosis of CSID

The standard method of confirming a diagnosis of suspected CSID is with an endoscopic biopsy, a tissue sample, taken from the small intestine for laboratory analysis. A sucrose breath hydrogen test has also been used, which is less invasive but more susceptible to error and can provoke GI symptoms due to the amount of sucrose that must be ingested to perform the study.

Developed more recently, a carbon-13 (¹³C-sucrose) breath test is noninvasive with a high degree of accuracy and avoids provoking symptoms. A genetic test of more common mutations in the CSID gene offers another less invasive simpler alternative.

In the absence of an early diagnosis, people with CSID may go for years without an accurate diagnosis. Usually they will exhibit normal growth, and often report persistent diarrhea. The diagnosis of CSID may be delayed while more common causes or disorders are investigated or diagnosed.

Treatments

People of any age need to consume the right nutrients in their diet to ensure proper nutrition. In a person with CSID, dietary restrictions often require life-long adherence to a strict sucrose-free diet. This can vary depending on symptoms, but foods high in sucrose should be avoided.

Foods high in a starch component (amylopectin) including cereals, breads, pastas, and potatoes may also need to be excluded, especially during the first years of life. Starch

tolerance is generally improved after the age of 3–4 years, and rice starch and maize starch are easier to digest.

Taking a small amount of baker's yeast along with sucrose-containing foods has been found to reduce symptoms. However, baker's yeast has an unpleasant taste.

An alternative to traditional baker's yeast is sacrosidase (Sucraid), a liquid preparation that is tasteless when mixed with water. Sacrosidase is an enzyme replacement therapy available by prescription. Sucraid was originally approved by the US Food and Drug Administration (FDA) in 1998 for treating CSID. Studies indicate its effectiveness in reducing symptoms while allowing a less restrictive diet and more normal lifestyle.

The Healthcare Team

Dietary management of malabsorption disorders like CSID can be challenging. Talk to your doctor and a registered dietician about what foods may cause digestion problems. Ask about alternatives. Learn how to read food labels and what to avoid. Be aware of possible sugar content of medicines, if needed. In children, challenges increase as they begin to exert more independence. As a parent or care provider you will want to work with your child's doctor to understand the condition and related limitations. This will provide you with the knowledge and ability to manage the child's symptoms and needs. At any age, working together with your healthcare providers will help ensure that proper nutrition is maintained while at the same time keeping symptoms under control.

***Adapted from the following primary sources**

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