Congenital Sucrase-Isomaltase Deficiency

What We Know

› Congenital sucrase-isomaltase deficiency (CSID) is a rare, autosomal recessive, genetic disorder in which deficiency or absence of the enzymes sucrase and isomaltase result in malabsorption of sucrose (i.e., granulated white table sugar) and maltose from starch sources that are high in amylopectin (e.g., wheat, potatoes, corn sweeteners). Signs and symptoms (e.g., diarrhea, abdominal pain) occur when sucrose is consumed, and vary in intensity according to the degree of sucrase-isomaltase deficiency as well as the amount of sucrose or starch consumed. CSID is frequently misdiagnosed as intolerance to cow’s milk protein, colitis, irritable bowel syndrome (IBS), chronic diarrhea, or food allergies. Treatment options include the restriction of dietary sucrose based on the severity of symptoms and the use of the artificial enzyme sacrosidase (Sucraid oral solution) to assist digestion of sucrose (1-2,3,4,5,6,7).

› Signs and symptoms of CSID (1,3,5,7):

• Manifestations of CSID vary depending on the degree of sucrase-isomaltase deficiency and the amount of sucrose or starch consumed. Potential signs and symptoms include the following:
  – Chronic watery or acidic diarrhea
  – Colitis
  – Weight loss
  – Vomiting
  – Abdominal pain and distension
  – IBS
  – Colic
  – Dehydration
  – Malnutrition
  – Slowed growth
  – Jaundice
  – Upper respiratory infection
  – Frequent viral infections
  – Failure to thrive

› Risk factors for CSID (4,5,7):

• CSID is an autosomal recessive condition, meaning both parents must contribute the defective gene in order for the child to have CSID. Carriers can also exhibit signs and symptoms of CSID, but they do so to a lesser degree.

• CSID occurs most frequently in persons living in or from the following countries/regions:
  – England
  – Germany
  – Ireland
  – Scandinavia
  – Eastern Europe
  – Russia
  – Iceland
  – Baltic states
Treatment of CSID: Treatment is focused on preventing signs and symptoms of CSID by instituting a diet that is restricted in sources of sucrose and high in amylpectin starch. Common foods that are high in sucrose and amylpectin include the following:
- All forms of table sugar and foods made with table sugar
- Potatoes
- Corn-sweeteners
- Use of the artificial enzyme sacrosidase can assist in sucrose digestion but does not aid in maltose digestion; persons receiving sacrosidase require starch restriction if symptoms persist.

Recent research findings on CSID and diet:
- No current research regarding CSID is published.

What We Can Do:
- Become knowledgeable about CSID and diet so you can accurately assess your patients’ personal characteristics and health education needs; share this information with your colleagues.
- Educate your patients regarding the importance of reporting any health-related changes to the treating clinician.
- Assess your patients and their family members for knowledge deficits about the prescribed treatment regimen, and emphasize the importance of strict adherence to the plan of care and continued medical surveillance to monitor health status.

Note:
- Recent review of the literature has found no updated research evidence on this topic since previous publication on February 20, 2015.

Coding Matrix:

References: