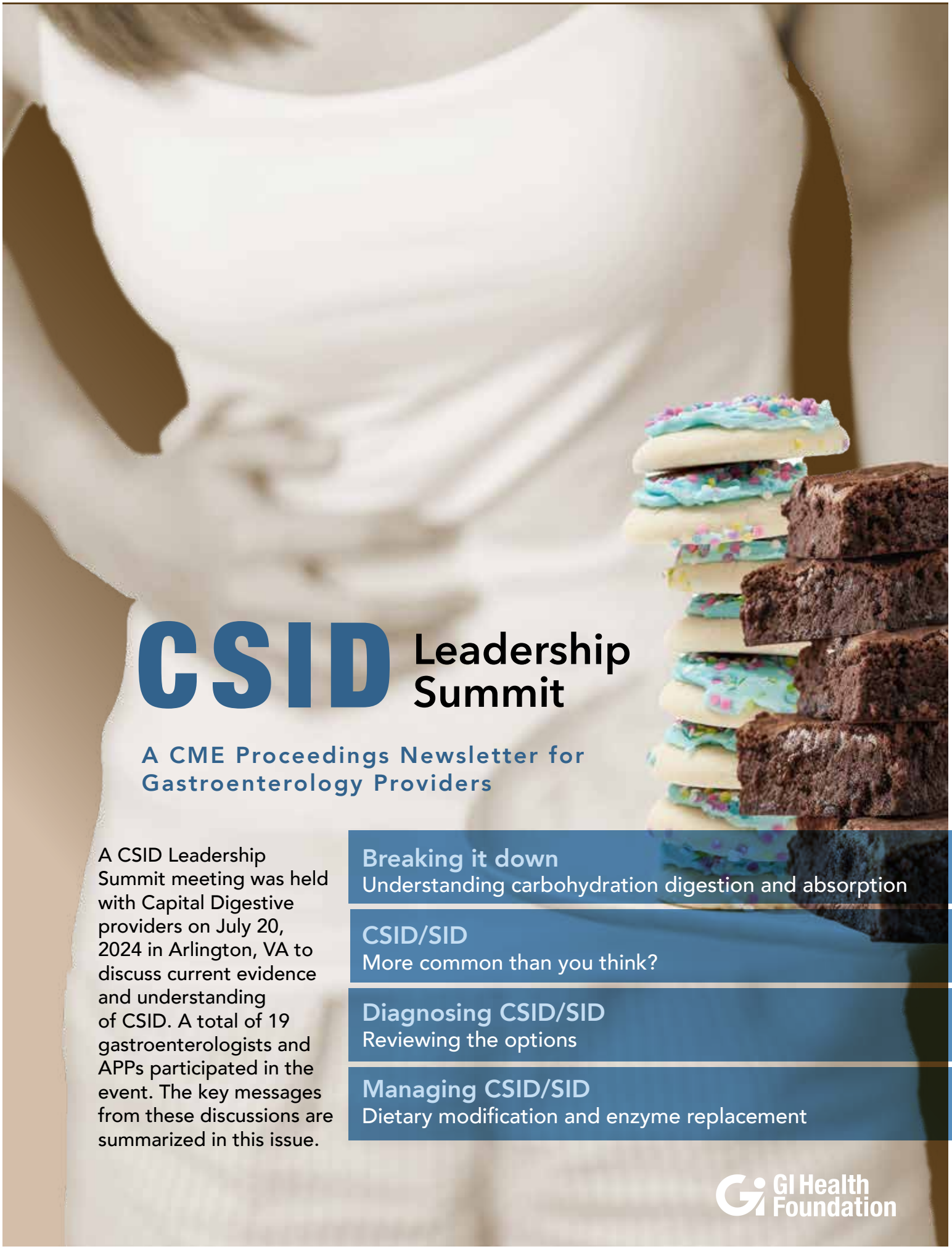

CSID Leadership Summit

A CME Proceedings Newsletter for Gastroenterology Providers

A CSID Leadership Summit meeting was held with Capital Digestive providers on July 20, 2024 in Arlington, VA to discuss current evidence and understanding of CSID. A total of 19 gastroenterologists and APPs participated in the event. The key messages from these discussions are summarized in this issue.

Breaking it down
Understanding carbohydration digestion and absorption

CSID/SID
More common than you think?

Diagnosing CSID/SID
Reviewing the options

Managing CSID/SID
Dietary modification and enzyme replacement

GI Health Foundation

2024 Mid-Atlantic Conference CSID APP Leadership Summit - A CME Proceedings Newsletter for Advanced Practice Providers

To claim 1.0 credit hour for this activity, please visit:
https://education.gihealthfoundation.org/content/capital-digestive-csid-leadership-summit-cme-proceedings-newsletter-advanced-practice

Release date: July 11, 2025
Expiration date: July 11, 2026

Target Audience

The joint providership estimates the initiative will attract an audience of 5,000 participants, including physicians, nurse practitioners, nurses, and physician assistants. A comprehensive reach optimization effort will be driven by the Gi Health Foundation (GiHF) and Gastroenterology & Hepatology Advanced Practice Providers (GHAPP) proprietary databases and educational portals. Outreach will focus on health care professionals who have opted-in for educational updates from GiHF and GHAPP.

Program Overview

The joint providership of Medical Education Resources (MER) and GiHF proposes to develop an accredited proceedings e-Newsletter on the management of patients with CSID. In addition to the content discussed during the CSID leadership summits and virtual mentorship sessions, additional expert opinions, recent publications, abstracts, and presentations from congress meetings will be reviewed as part of the e-Newsletter development process. The e-Newsletter will launch in February 2024 and reach more than 5,000 participants via GiHF's and GHAPP's educational portals and will be posted for a period of at least 12 months.

Educational Objectives

Upon completion of this activity, participants should be able to:

1. Describe the prevalence of CSID in patients with common GI disorders
2. Incorporate current diagnostic strategies to differentiate CSID from other causes of persistent diarrhea seen in clinical practice, particularly among patients with suspected IBS
3. Summarize benefits and limitations of current treatment

Faculty and Planners

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Accreditation:

JOINTLY ACCREDITED PROVIDER™

In support of improving patient care, this activity has been planned and implemented by Medical Education Resources (MER) and GI Health Foundation (GIHF). MER is jointly accredited by the Accreditation Council for Continuing Medical Education (ACCME), the Accreditation Council for Pharmacy Education (ACPE), and the American Nurses Credentialing Center (ANCC), to provide continuing education for the healthcare team.

Physician Credit:

Medical Education Resources estimates each educational activity for a maximum of **1.0** *AMA PRA Category 1 Credit*™.

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MER and GIHF planners have no relevant financial relationships to disclose.

Disclosures:

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- Consultant
  - AbbVie, Boehringer Ingelheim, GlaxoSmithKline, Intercept, Ipsen, Madrigal, Novo-Nordisk, Salix

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Method of Participation:

There are no fees for participating in and receiving credit for this activity. During the period July 11, 2025 - July 11, 2026 participants must 1) read the learning objectives and faculty disclosures, 2) study the educational activity, 3) complete the posttest by recording the best answer to each question, and 4) complete the evaluation form. A statement of credit will be issued only upon receipt of a completed activity evaluation form and a completed posttest with a score of 75% or better. Please visit https://education.gihealthfoundation.org/content/capital-digestive-csid-leadership-summit-cme-proceedings-newsletter-advanced-practice complete the post-test and evaluation.

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MEDICAL EDUCATION RESOURCES

GI Health Foundation

Supported by an educational grant from QOL Medical LLC.

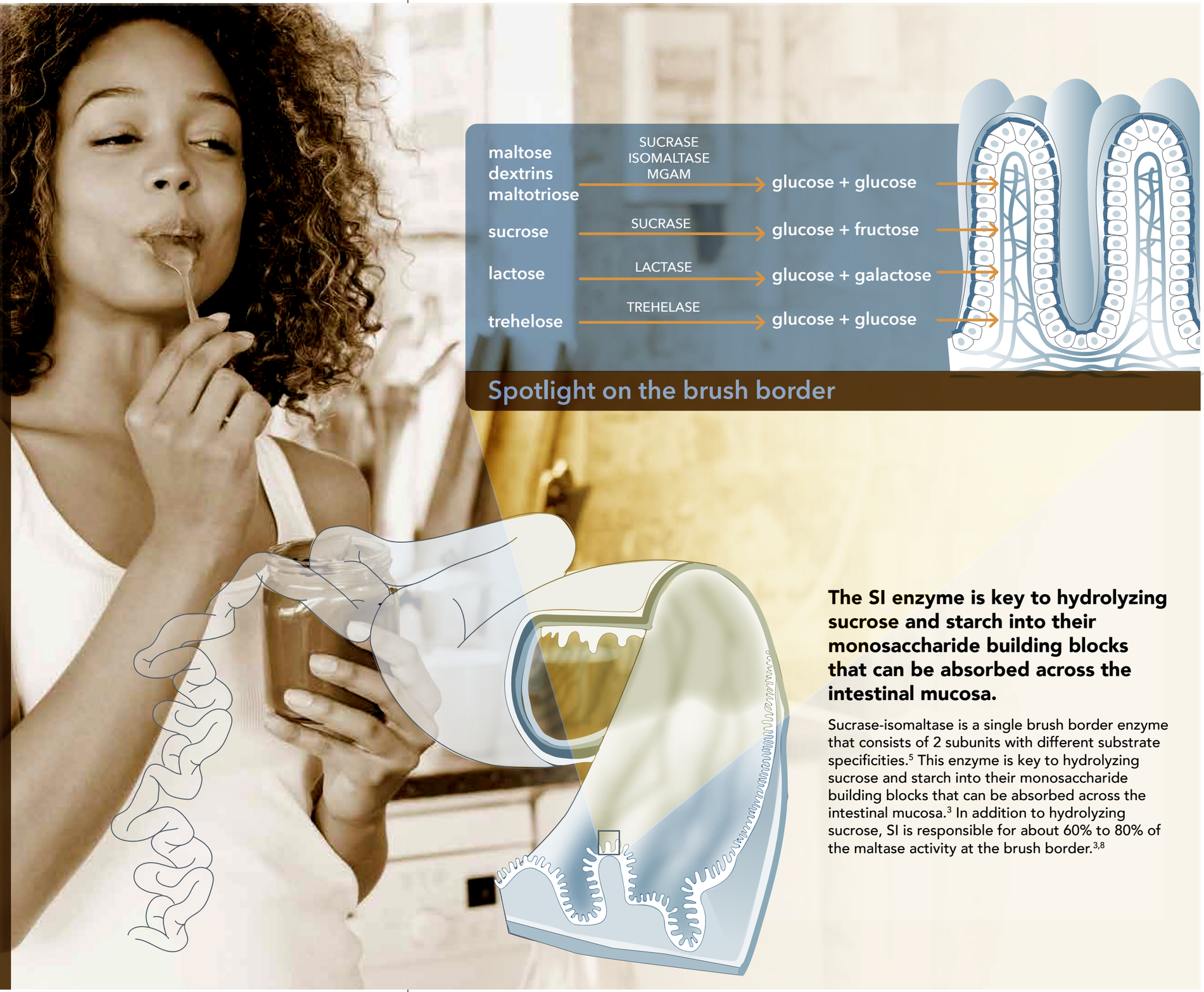
BREAKING IT DOWN

understanding carbohydrate digestion and absorption

Carbohydrates make up nearly half of the average Western diet[1] and are the most important source of food energy among the macronutrients.[2]

Most digestible dietary carbohydrates are table sugars (sucrose) and plant starches that are composed of different α-linked sugars.[3] Because sugar transporters in the small intestine can only transport monosaccharides (ie, simple sugars), all products of carbohydrate digestion must be reduced to monosaccharides for absorption across the intestinal epithelium.[4] This process begins in the mouth with salivary α-amylases that hydrolyze starches into smaller polysaccharides and α-dextrins (starch fragments).[2,4,5] As food passes from the stomach to the small intestine, pancreatic amylases are released to resume the digestion of starch into smaller di- and trisaccharides.

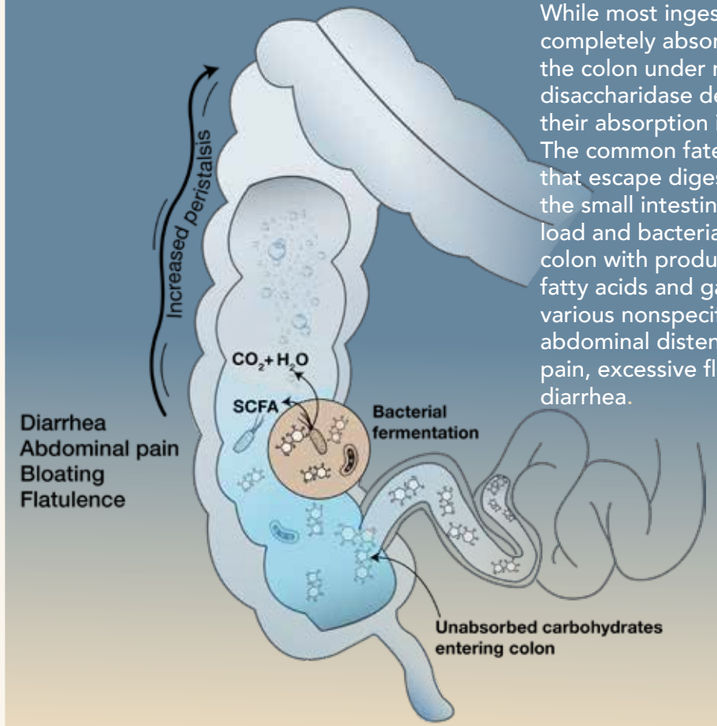
The next and final phase of carbohydrate digestion is taken over by enzyme complexes bound to the microvilli of the enterocytes lining the small intestine that constitute the intestinal brush border.[6] With up to 1000 microvilli and numerous folds to increase their surface area, these enterocytes are specially equipped to maximize absorption.[7] Here at the brush border, a group of enzymes, or disaccharidases, hydrolyze disaccharides into their monosaccharide components so they can be transported across the intestinal epithelium into the portal circulation.[6,7] The main disaccharidases responsible for this phase of digestion are glucoamylase, sucrase-isomaltase (SI), and lactase.



The SI enzyme is key to hydrolyzing sucrose and starch into their monosaccharide building blocks that can be absorbed across the intestinal mucosa.

Sucrase-isomaltase is a single brush border enzyme that consists of 2 subunits with different substrate specificities.[5] This enzyme is key to hydrolyzing sucrose and starch into their monosaccharide building blocks that can be absorbed across the intestinal mucosa.[3] In addition to hydrolyzing sucrose, SI is responsible for about 60% to 80% of the maltase activity at the brush border.[3,8]

CSID from malabsorption to symptoms



While most ingested carbohydrates are completely absorbed before reaching the colon under normal conditions, disaccharidase deficiencies can impair their absorption in the small intestine.[6] The common fate of any carbohydrates that escape digestion and absorption in the small intestine is increased osmotic load and bacterial fermentation in the colon with production of short-chain fatty acids and gases.[4,12] This can lead to various nonspecific symptoms such as abdominal distension, bloating, cramping, pain, excessive flatulence, and/or osmotic diarrhea.

> **The downstream effects of bacterial fermentation in the colon can generate IBS-like symptoms—not necessary abdominal pain with altered bowel habit, but can simply be bloating, dyspepsia, flatulence, or abdominal discomfort.**
>
> Brooks Cash, MD

what's in a name?

Although intolerance, malabsorption, maldigestion, and enzyme deficiency are often used interchangeably,[4] these terms describe different aspects of pathogenic processes, with different diagnostic and potentially different therapeutic implications.[12] Carbohydrate malabsorption generally refers to incomplete absorption of carbohydrates in the small intestine, leading to undigested carbohydrates reaching the colon.[12] However, carbohydrate malabsorption may not be pathological and is considered clinically relevant only when it results in abdominal symptoms (ie, intolerance).[12]

Sucrase - isomaltase

deficiency
Low expression of functioning SI enzyme in the small intestine

malabsorption or maldigestion
Incomplete absorption of sucrose and isomaltose in the small intestine, leading to undigested carbohydrates reaching the colon[12]

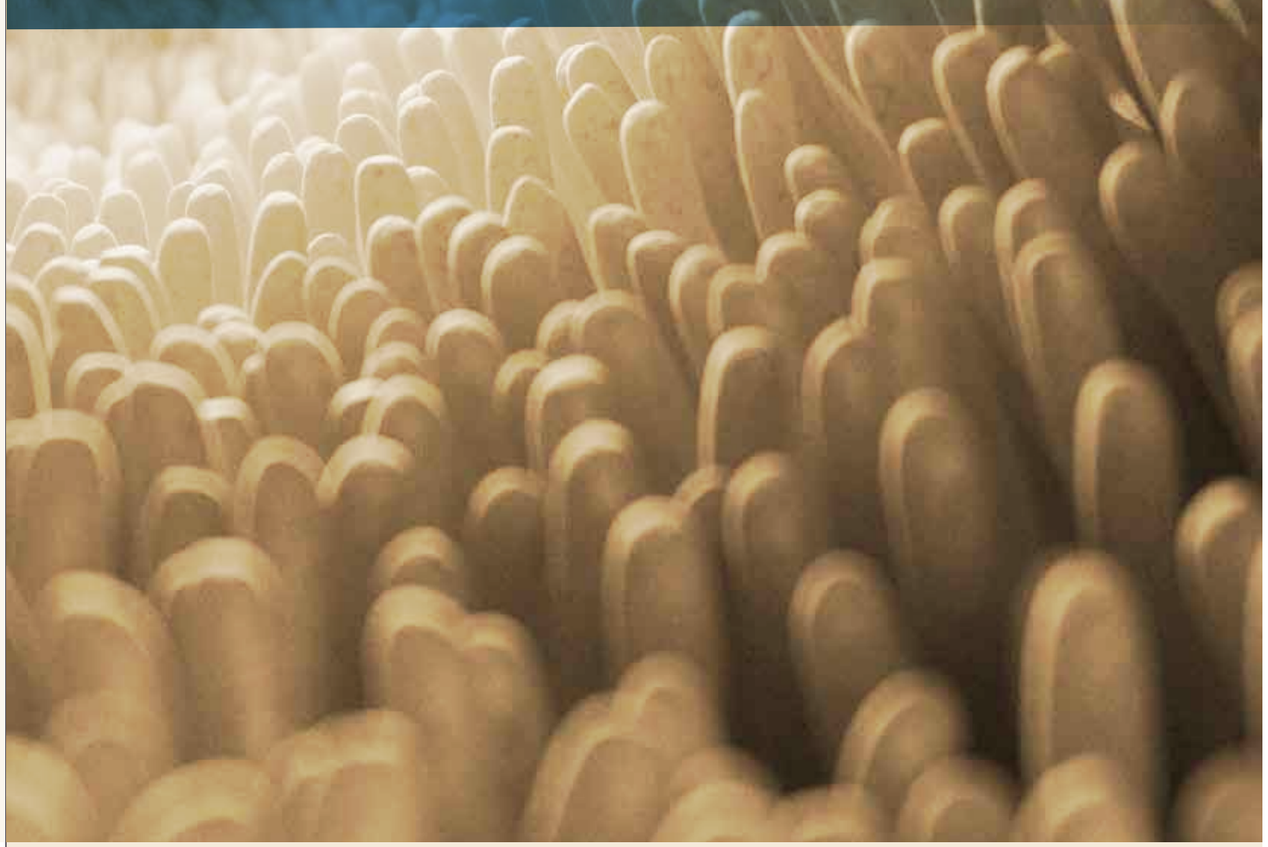
intolerance
GI symptoms after sucrose and/or starch ingestion due to sucrose and/or isomaltose malabsorption

Congenital sucrase-isomaltase deficiency (CSID) results when patients inherit defective copies of the sucrase-isomaltase (*SI*) gene due to either recessive homozygous or compound heterozygous mutations that reduce or abolish enzymatic activity.[9] Although numerous mutations in the *SI* genes have been identified, 4 variants account for many of the clinical symptoms associated with CSID.[10]

Acquired or secondary forms of sucrase-isomaltase deficiency can also occur in patients with chronic diarrhea from other causes such as villous atrophy or alteration, infection, and rapid transit.[11] The clinical impact of sucrase-isomaltase deficiency in these disorders may be transient, with enzymatic activity returning to normal with resolution of the underlying disorder.[11] The faculty noted that although pediatric gastroenterologists may be more likely to encounter congenital deficiencies, they suspect that most cases of sucrase isomaltase deficiency they see in their adult patients represent acquired deficiencies.

Secondary causes of sucrase-isomaltase deficiency[3,4]

- Pancreatic insufficiency or resection
- Age-related decrease in lactase activity
- Mucosal disease (eg, celiac disease, Crohn's disease)
- Loss of mucosal surface area (intestinal resection or bypass)
- Small intestinal bacterial overgrowth (SIBO)
- Radiation injury
- Drug/toxin injury (eg, antineoplastic agents, neomycin)
- Infection (HIV, Giardiasis, rotavirus, *Yersinia enterocolitica*)



recognizing sucrase-isomaltase deficiency

Although symptoms usually appear early in life, the clinical presentation and severity of CSID/SID vary depending on the nature and position of the SI mutations, as well as their homozygous or heterozygous combinations.[9,13] Accordingly, sucrase activity in patients with CSID/SID can range from completely absent to low residual activity, while isomaltase activity can range from absent to normal.[8] Maltase activity is also reduced significantly in most patients with CSID/SID.[8,14] In addition to residual enzyme activity, many other factors can affect the onset and symptoms of CSID/SID, including the amount of dietary sugar and starch consumed, the rate of gastric emptying, activity of other intestinal disaccharidases, and the metabolic activity of fermenting bacteria.[3,8,11]

In contrast to the classic, severe presentation of CSID in patients with homozygous *SI* mutations,[8] a broad range of phenotypes has been observed in adults with CSID/SID. Many of the symptoms of CSID overlap with those of IBS, particularly IBS-D.[10] Like IBS, CSID/SID symptoms often occur postprandially, but patients with the condition may be likely to associate symptoms with sweets and other high-sucrose foods. Patients with CSID/SID may report a lifelong history of symptoms, potentially with avoidance of carbohydrates or sweet foods, as well as family members with similar symptoms.

The clinical presentation of CSID/SID can vary widely based on residual SI activity, dietary sucrose and starch consumption, and patient age.[3,11]

Clinical clues
to sucrase-isomaltase deficiency[15,16]

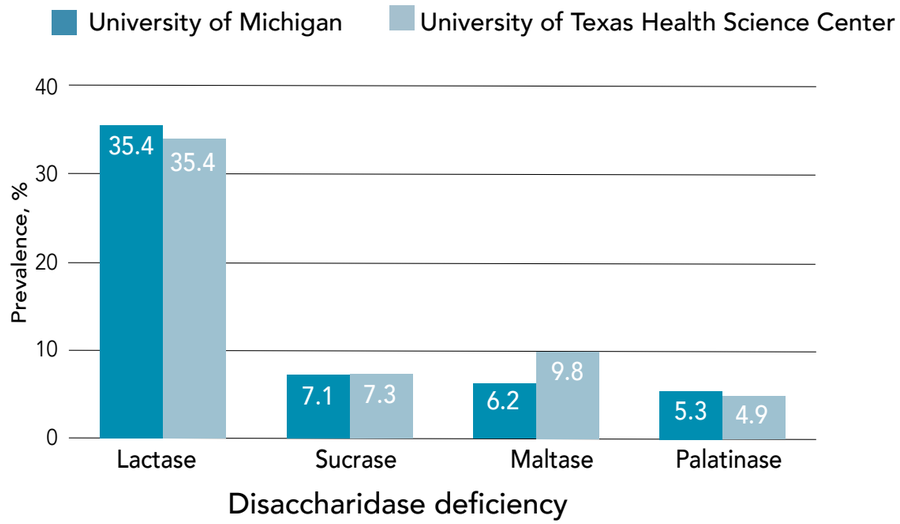
- Lifelong symptoms
- Postprandial symptoms
- History of avoiding sweets
- Family members with similar symptoms
- No evidence of alarm features

CSID/SID
more common than you think?

Although once believed to be a rare autosomal recessive disorder,[8] both clinical and genetic data indicate that CSID is more common than previously believed.[13,17-20] This is increasingly apparent in patients with unexplained functional GI symptoms, particularly with presumed IBS. Studies in adults suggest that many patients with CSID are diagnosed with IBS at some point in their lives.[10] An interim analysis of 154 adults meeting Rome IV criteria for IBS-D or functional diarrhea found that 1 in 14 (7.14%) symptomatic patients had sucrase and maltase deficiencies on disaccharidase analysis.[17] Previous studies have also reported a high prevalence of CSID in patients with chronic unexplained GI symptoms.[21,22]

Growing evidence also suggests that specific pathogenic *SI* gene variants are more common in patients with IBS than those without.[9,13] In a study involving 1031 patients with IBS, patients with IBS were nearly twice as likely to have a genetic *SI* mutation compared with controls (odds ratio=184).[9] In a larger study involving 2207 patients with IBS, 4.2% of patients with IBS-D were found to carry rare *SI* pathogenic variants, a higher frequency relative to a large matched reference population.[13]

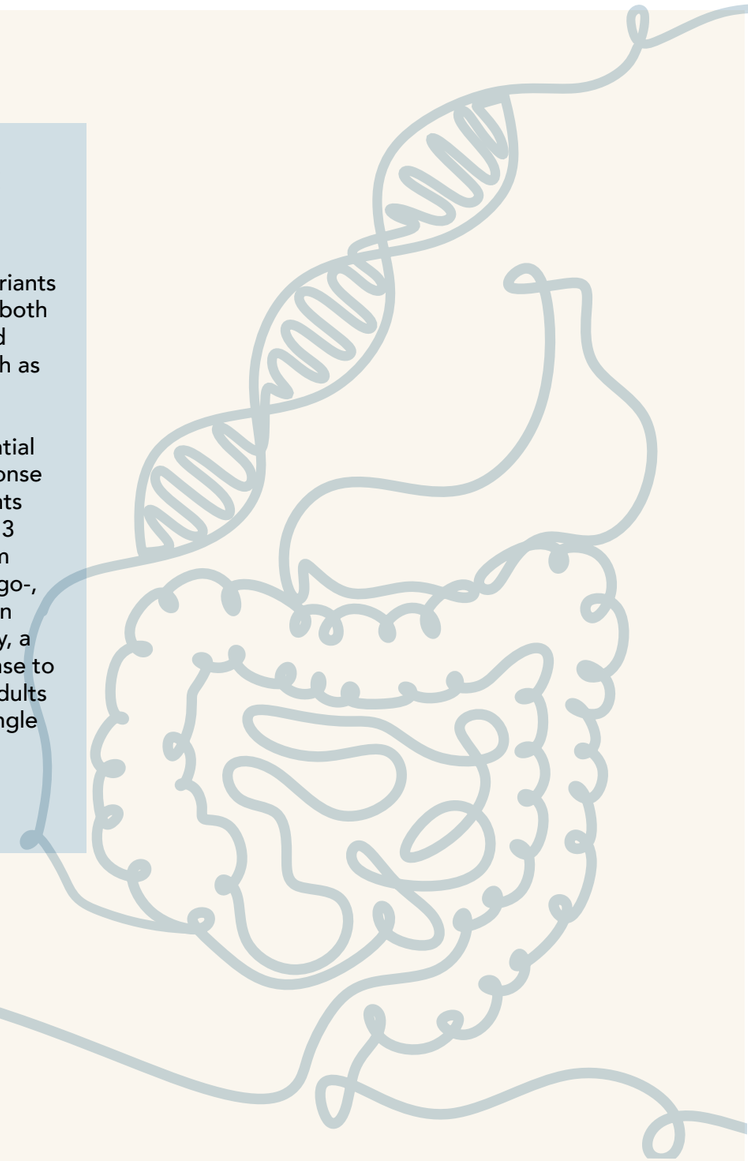
Prevalence of disaccharidase deficiency in adults with Rome IV-defined IBS-D or functional diarrhea (N=154)[11]



what does a pathogenic *SI* variant mean?

The clinical implications of pathogenic SI variants continue to be explored. Several studies in both pediatric and adult patients have associated such variants with chronic GI symptoms such as diarrhea and abdominal pain.[13,18,23]

Other data are emerging regarding a potential association between *SI* mutations and response to dietary modification. In one study, patients with IBS-D and pathogenic SI variants were 3 to 4 times less likely to experience symptom relief with a low FODMAP (fermentable, oligo-, di-, mono-saccharides, and polyols) diet than patients without such variants.[24] Additionally, a pilot study has demonstrated better response to a starch and sucrose-reduced diet among adults carrying 2 *SI* variants than those carrying single or no variants.[25]



The point here is not the absolute numbers. The point is that this is present in our patients when we actually look for it.

Brooks Cash, MD

diagnosing CSID/SID

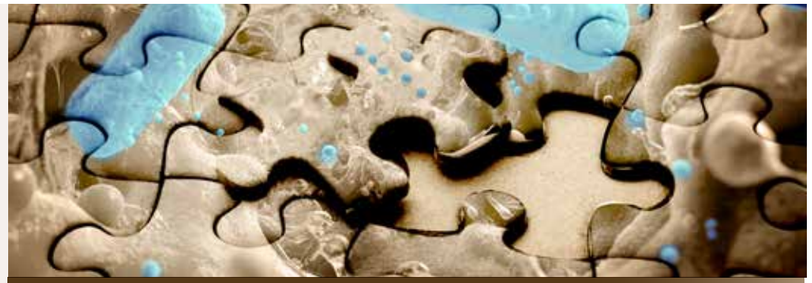
Endoscopic small bowel biopsies assayed for disaccharidase (lactase, sucrase, isomaltase, and maltase) activities are the gold standard for diagnosing intestinal disorders associated with carbohydrate metabolism.[3,8] Key advantages of these tests is that they allow for the evaluation of all disaccharidases and can help distinguish between congenital and secondary deficiencies. However, assay results vary considerably as their accuracy depends on proper specimen collection and handling.[20]

procedure matters

1 **COLLECT.** First biopsies should be obtained from the distal duodenum or proximal jejunum and the samples placed in a empty eppendorf tube. Do not place the tissue on gauze, filter paper, or use any type of support medium, not even saline.

2 **FREEZE.** Place eppendorf tube with collected sample immediately on dry or wet ice and freeze within 2 hours of collection at -20° C to -70° C.

3 **SHIP.** Samples should be shipped frozen on dry ice to appropriate lab promptly on the day of collection.



There is little evidence to inform the positioning of testing for SIBO vs SID in the diagnostic algorithm for patients with IBS-like symptoms. The faculty noted that they sometimes treat patients empirically with a nonabsorbable antibiotic for dysbiosis before evaluating for SID. This approach is based on speculation that SIBO is more common in this population, recognition that SIBO can lead to acquired SID, and poor confidence in the accuracy of breath testing. However, they may be more likely to evaluate for SID in those with chronic diarrhea and symptoms that are suggestive of SID, such as postprandial distress and bloating. While these are reasonable approaches, the faculty emphasized that there is likely more than one perturbation in patients with IBS, and that **SIBO and SID are individual pieces of a more complex puzzle**.

non-invasive tests

Hydrogen-methane breath tests measure exhaled hydrogen levels produced by bacterial fermentation of a test carbohydrate.[11] Although these tests are simple and can be performed by patients at home, they cannot differentiate between small intestinal bacterial overgrowth (SIBO) and CSID.[26] Further, the 50-g required sucrose load can cause significant symptoms for patients with CSID.

The **^{13}C-sucrose breath test** is a more direct measure of sucrase activity[27] that can be stocked in the office or sent directly to patients. Although this test requires fewer pre-test restrictions than the sucrose-hydrogen-methane test, it has not been validated for use in clinical practice.[10] Overall, the limitations of breath testing have been well recognized and their role in evaluating patients with IBS-like symptoms continues to be debated.[4,26,28,29]

A 4-4-4 sucrose challenge is a simple test that consists of monitoring for the presence of symptoms for a 4 to 8-hour period after the patient drinks 4 ounces of water with 4 tablespoons of dissolved table sugar.[11,30] A variation of this method was recently evaluated in an in-home study in 45 patients with confirmed CSID and 118 healthy controls.[31] After an overnight fast, patients ingested 50 g of sucrose dissolved in water and self-reported severity of 6 GI symptoms on a 10-point scale through 4 hours. When optimized by gender, a worsening in global symptoms score at 1 and 2 hours was found to have an 87% sensitivity and 81% specificity for identifying CSID cases. Although more data are needed to further characterize and validate this test, these findings suggests that the sucrose challenge symptoms test could serve as a practical diagnostic tool to help identify patients with CSID.

Mutations in the *SI* gene causing CSID can be identified by **genetic testing** using saliva or blood.[3] Although a positive test can confirm a diagnosis of CSID, a negative test does not rule out the condition as the available tests identify only a small number of *SI* mutations.

Managing CSID/SID
with dietary modification

Enzyme replacement therapy and dietary modification of starch and sucrose are the cornerstones of CSID management.[3,10] Recognizing that the degree of sucrose and starch intolerance can vary among patients, dietary modification is accomplished on a trial and error basis, adjusting specific foods as needed based on symptoms.[15] Although this process can be initiated by restricting both sucrose and starch, the faculty shared that they more commonly begin by restricting sugar only and follow with starch reduction if symptoms persist. However, starch reduction may be considered more important in patients who are very symptomatic.

The process of dietary modification can be complex, involving several weeks of elimination of dietary sucrose and starch, followed by gradual reintroduction of foods into the diet.[10] Additionally, recognizing the increasing prevalence of disordered eating in patients with DGBIs (Disorders of Gut-Brain Interaction),[33-35] it is essential to work with patients to determine the least restrictive diet that is tolerable. With these considerations in mind, the faculty emphasized the importance of engaging a dietitian to help patients with this process. In addition to working with patients to determine their individual tolerance of sucrose- and starch-containing foods, dietitians can teach patients to understand food labels so they better recognize sucrose and starch in foods.[36] For example, patients who are modifying their starch intake should be taught to recognize ingredients such as dextrins, maltodextrins, and glucose polymers as starches.

Although dietary restriction can be effective, follow-up studies indicate that only a minority of patients remain consistently asymptomatic with this approach, with up to 75% of patients continuing to experience diarrhea, gas, and/or abdominal pain. Further, only half of children are typically compliant with the prescribed diet.[8,37,38]

Foods that are low in sucrose and starch[30]

DAIRY[a]
Cow's milk
Cream cheese
Half and half
Hard cheeses (cheddar, colby, mozzarella, swiss, parmesan, provolone)
Plain cottage cheese
Plain yogurt sweetened with fructose or dextrose
Ricotta cheese
Sour cream
Whipping cream

PROTEIN[b]
Beef
Chicken
Eggs
Fish
Lamb
Pork
Tofu
Turkey

NUTS & SEEDS[c]
Almonds
Almond butter
Brazil nuts
Flax seeds
Hazelnuts
Macadamia nuts
Peanuts
Peanut butter
Pecans
Pumpkin seeds
Sesame butter (tahini)
Walnuts

VEGETABLES
Alfalfa sprouts
Artichoke
Asparagus[d]
Bamboo shoots
Bok choy
Broccoli
Brussels sprouts[d]
Cabbage[d]
Cauliflower[d]
Celery
Cucumber
Eggplant
Green beans
Greens (collards, kale, mustard, turnip, and chard)
Lettuce (arugula, endive iceberg, romaine)
Mung bean sprouts
Mushrooms
Peppers (red, green, and yellow)
Radishes
Rutabaga
Spaghetti squash
Spinach
Tomatoes
Turnips
Yellow squash
Zucchini

FATS
Any vegetable oils
Butter

FRUITS
Avocado
Blackberries
Blueberries
Cherries
Cranberries
Currants
Figs
Grapes
Kiwi
Lemons
Limes
Olives
Papaya
Pomegranate
Prunes
Raspberries
Rhubarb
Strawberries

[a]Full-fat dairy products may be used if more calories are indicated. Avoid processed cheeses or cheese products that contain sucrose or starch fillers. If lactose intolerant, avoid dairy foods. Substitute lactose-free milk, unsweetened almond milk, or soy milk for cow's milk.
[b]Avoid processed meats such as bacon, sausage, luncheon meat, paté, and liverwurst that are cured with sucrose or have starch fillers.
[c]Nuts and seeds can be difficult to digest in general. Most nuts and seeds contain varying amounts of sucrose and starch. When starting the diet, it is best to avoid nuts and seeds the first two weeks. It is important to keep the portion size small (in general a serving is <1 ounce for nuts).
[d]These vegetables may cause gas in all individuals and should be monitored closely.

Managing CSID

with enzyme replacement

Treatment of CSID with enzyme replacement therapy can improve symptoms while allowing patients to consume a more liberal diet.[8,10,21] Sacrosidase, which is sucrase enzyme derived from *Saccharomyces cerevisiae*, was approved by the FDA for treatment of CSID in 1998.[20] This product is usually taken with each meal or snack, mixed in with 2 to 4 ounces of milk, water, or formula.[40] While sacrosidase aids in sucrose digestion, starch restriction may still be needed for some patients as it does replace deficient isomaltase.[30]

In a randomized, double-blind trial, 81% of patients using full-strength sacrosidase were able to remain asymptomatic while consuming an unrestricted diet compared with 78% untreated during the baseline, diet-restricted period.[8,39] More recently, a chart review of 258 adults with chronic unexplained GI symptoms demonstrated that dietary counseling and/or enzyme replacement improved symptoms in the 60% of patients who had positive breath tests for sucrose malabsorption.[21] When discussing these data, the faculty shared that they typically see symptom improvement once their patients initiate sacrosidase treatment rather than complete symptom resolution. Sacrosidase is well tolerated, with the most common adverse events reported being constipation, headaches, and sleep disturbances.

Although sacrosidase is the only approved treatment for CSID, nonprescription enzyme blends are available.[20] For example, a combination of invertase and glucoamylase is available in a capsule formulation that is intended for administration immediately before or with the first bite of each meal.[41]

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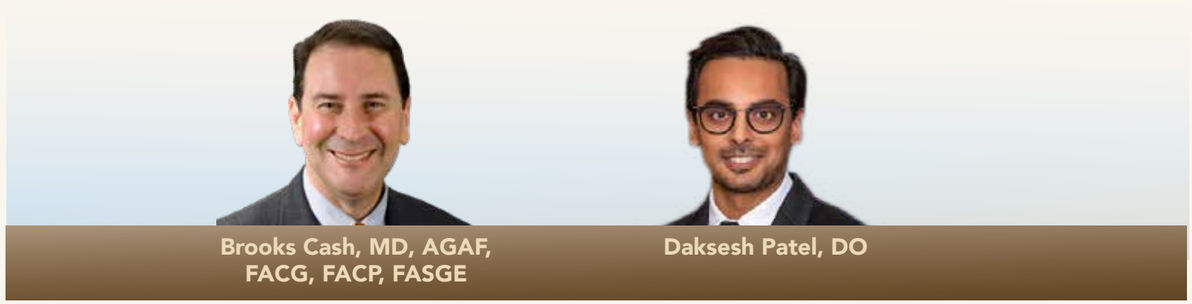
Faculty



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