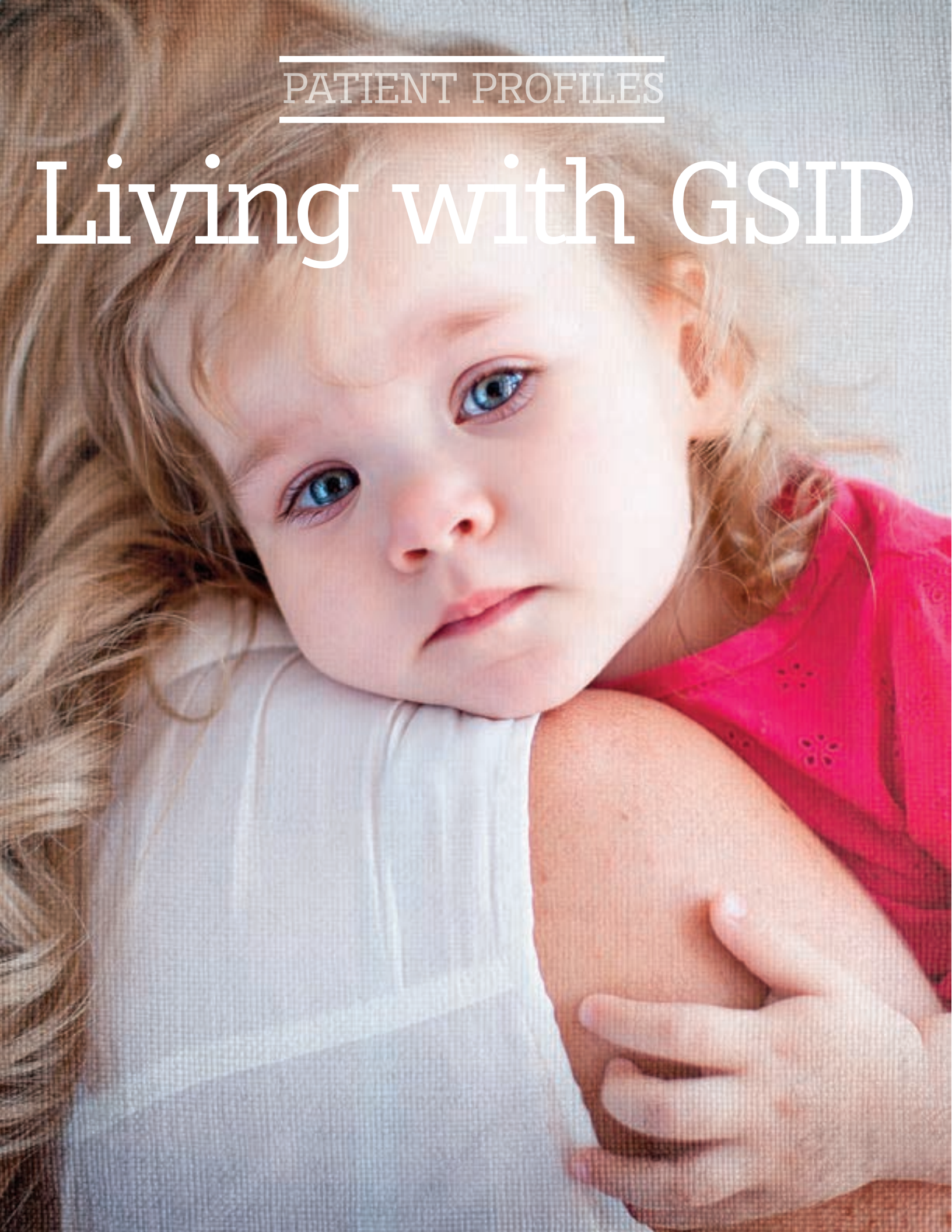
PATIENT PROFILES

Living with GSID



Jessica

Jessica, a 4-month-old baby girl, was admitted to the hospital through the ER after experiencing chronic diarrhea and continual weight loss over the past month.

Jessica was a full-term healthy baby weighing 7 pounds. Initially Jessica breastfed, and did not display any bouts with diarrhea or weight loss. Jessica's mother switched to formula after weeks of breastfeeding. Once beginning formula, Jessica started experiencing "watery" diarrhea 4 to 5 times a day with occasional solid stools. Even after experimenting with lactose-free formulas and amino-based formula, there was no relief from Jessica's chronic diarrhea.

Upon physical exam, the doctors noted that all Jessica's developmental milestones had been met. She was alert and crying and still had a healthy appetite of 4 to 5 bottles per day. An abdominal exam revealed distention and facial grimacing when pressure was administered to her abdominal region. At 4 months old Jessica's weight fell below normal for her age and her skin was dry and pallor. Blood test levels were within normal limits, and stool cultures for bacteria and parasites were negative. Results from an endoscopy, colonoscopy and histology were unremarkable.

Diagnosis

Jessica's symptoms led physicians to believe she was suffering from a genetic sucrase-isomaltase deficiency. Symptoms for this genetic deficiency typically reveal themselves after weaning a baby and beginning formula consumption.

A sucrase deficiency leads to a buildup of luminal fluid and bacteria, which causes a fermentation process and generation of gasses. The digestive symptoms include: abdominal cramps, bloating, excess gas production, vomiting and chronic diarrhea. As a result of this deficiency, patients affected have difficulty gaining weight and may become malnourished.

The diagnosis of genetic sucrase-isomaltase deficiency can be challenging because many of the symptoms are non-specific. Patients are more likely to be diagnosed with more common causes of diarrhea such as toddler's diarrhea, lactose intolerance or celiac disease.

Jessica continues to be treated for genetic sucrase-isomaltase deficiency and is on the road to recovery. **GSID**

Knowledge is Power

The first thing to realize is that knowledge is power. Just as you, the parent or caregiver, will feel better equipped to handle the diagnosis as you learn more, so will your child. The more information you and your child can learn, the better you and your child will feel about your family's ability to deal with GSID.

One Mother's Story

"At first my daughter was vomiting a couple times per week, then it got progressively worse over the next nine months to the point of her vomiting 5-10 times per day, 10-15 bowel movements (ranging from clay-like, fatty, or liquid diarrhea) per day, severe abdominal distention, pain to the point of her being unable to sleep for periods longer than 30 minutes at a time without waking up screaming and clutching her stomach...and an extremely decreased appetite (not eating for 2-3 days at a time) with gastroparesis. She was emaciated (her weight was down to 16lbs. at 2 years of age) with noted muscle wasting, and she was scheduled for a PEG tube insertion until we found out about GSID. She tested positive and we immediately began taking the necessary steps toward treatment. She is now up to 22lbs. and no longer has any of her previous symptoms!" *–Mother of child diagnosed with GSID*

Michael

After 7 months of chronic explosive diarrhea, 19-month-old Michael was referred to a gastroenterologist. Adopted at 1 month old, Michael's medical history was unknown.

Michael has experienced "watery" and brown diarrhea 4 to 20 times a day. He has also suffered from weight loss, dehydration and excoriated buttocks (red, raw, broken down skin). Two months ago he was diagnosed with gastroesophageal reflux disease (GERD) and 2 weeks ago with toddler's diarrhea. Michael has been treated with antibiotics, a probiotic supplement, and pancreatic enzymes. These treatments have provided no relief or improvements for him. He is currently not taking any medication. Blood test levels were within normal limits, abdominal x-rays and ultrasound were unremarkable. Stool study revealed low pH, but no parasites. And Michael has no history of prior surgeries.

Diagnosis

Doctors suggested that Michael receive an esophagogastroduodenoscopy (EGD) with a disaccharidase assay. This procedure confirmed a diagnosis of genetic sucrase-isomaltase deficiency. Since being correctly diagnosed, Michael and his family are realizing great improvements in his symptoms. Michael is smiling again. **GSID**

Sarah

Sarah, a 9-year-old little girl, was brought to the doctor with abdominal pain, bloating, and excessive flatulence. She has been plagued with these symptoms for the past 4 years.

Sarah described her abdominal pain as intermittent with the pain increasing from dull to very sharp. She also experienced occasional vomiting and foul smelling diarrhea. After each meal, her abdominal pain would worsen and the pain lasted for several hours. As the day progressed Sarah's abdomen would become more distended, reaching a point where she looked "pregnant" in the evening. She often missed school because of her health problem.

Upon examination, it was noted that Sarah's abdomen was distended, and she experienced tenderness in her abdominal regions when pressed. Based on Sarah's age, her weight was just above the 25th percentile and her height was just below the 50th percentile. All her vital signs were all within normal limits. Sarah's mother said there is a family history of chronic abdominal pain on both sides. They have tried implementing a gluten free diet, a low FODMAP diet, and a lactose free diet. Unfortunately these diets didn't resolve any of Sarah's symptoms.

Diagnosis

After the examination, Sarah's doctor believed she had been suffering from a genetic sucrase-isomaltase deficiency. Sarah was given a sucrose intolerance hydrogen breath test that further aligned with the doctor's diagnosis. Sarah then underwent an upper endoscopy with disaccharidase analysis to definitively confirm she had been suffering from genetic sucrase-isomaltase deficiency. Sarah was treated by her doctor and her symptoms are now dissipating. **GSID**

Problem-solve with Your Child

Inevitably, your child will feel "different." This disorder deprives your child of the privacy of blending in for most social situations. If he/she is feeling frustrated, angry, or sad, help him/her express those emotions. Ask how he/she is feeling and always be available to offer a listening ear or a shoulder to cry on. Problem-solve with your child.

Lifetime Battle

Rebecca, “I cannot remember the last time I didn’t have symptoms.”

Rebecca

Rebecca, 36, has been plagued with chronic intermittent diarrhea, abdominal pain, and bloating since childhood. She cannot remember the last time she didn't have symptoms.

Rebecca is a 36-year-old white female patient who is married with 2 children. Since the age of 5, Rebecca has suffered with "stomach problems." These problems have included chronic intermittent diarrhea, abdominal pain, and bloating. As a child, Rebecca would miss school and these absences continued into her adulthood with missed days of work. Over the years, Rebecca has been hospitalized for dehydration and malnutrition. She noted that when she is exhausted her symptoms are more severe.

Rebecca has searched for a definitive diagnosis for her "stomach problems." She has visited a multitude of specialists who have diagnosed her with a plethora of conditions such as: von Willebrand's factor disease, esophagitis, gastritis, food allergies (soy, avocado, milk protein), gastroesophageal reflux disease, chronic diarrhea, irritable bowel syndrome, lactose intolerance, and hypertension. Unfortunately, no therapy prescribed for these conditions has provided any substantial benefit. Rebecca has noted that a strict vegetarian diet including fish does reduce her symptoms. She is currently taking hydrochlorothiazide, omeprazole, and a digestive enzyme supplement.

Providing a medical history prior to her examination, Rebecca stated that she never experienced fevers, nausea, vomiting, or bloody, black, or pale stools. When noting her family history, Rebecca revealed that her mother and maternal grandmother both have Crohn's disease. She had a C-section for one of her births and gallbladder surgery last year.

Diagnosis

With her chronic diarrhea, abdominal pain, and bloating, physicians performed an abdominal exam. Abdominal bloating and increased bowel sounds were noted during the exam. At 5 foot 4 inches Rebecca and weighing 94 lbs with a BMI of 16.23, Rebecca is medically underweight. Her vitals are unremarkable and her blood work and urine analysis were all within normal limits. An abdominal X-ray and abdominal ultrasound revealed no structural abnormalities.

Rebecca has been diagnosed with GSID. **GSID**

Vince

Vince is a 73-year-old male patient with early-stage Alzheimer's. He lives in a nursing home where he is cared for by his daughter and nursing staff. Vince has chronic non-specific GI symptoms including diarrhea, constipation, excessive flatulence, and bloating.

Vince is a 73-year-old male patient with early-stage Alzheimer's. He is a widow and has lived in a nursing home for the past 3 years where he is cared for by his daughter and nursing staff. Vince suffers from chronic non-specific GI symptoms including diarrhea, constipation, excessive flatulence, and bloating after every meal.

His nurse has noticed that his symptoms appear to be milder when abiding by a high protein and green vegetable diet, whereas most processed foods and foods that are high in sugar and carbs, including fruits cause an increase in his chronic symptoms.

Vince does not have symptoms like abdominal pain, fever, nausea, vomiting, or bloody, black, pale, or mucous stools. He has been diagnosed with colitis, irritable bowel syndrome, and gastroesophageal reflux disease. Although his family history is unknown, his medical history is available. Vince recalls that he has been suffering from diarrhea, flatulence, and bloating his whole life. His daughter added that "Dad would always have to go to the bathroom immediately after a meal."

Diagnosis

A physical exam was performed on Vince while he was sitting in an upright position. He is slightly overweight with blood pressure 143/89 , a heart rate of 49, and BMI at 28. The physician noted that Vince suffers from an excessive outward curvature of the spine, he also slouches and appears uncomfortable, constantly moaning while attempting to reposition himself. Upon conducting an abdominal exam, the physician observed increased bowel sounds, facial grimacing, and guarding on palpation of the epigastric region. No murmur, mass, fluid wave, rebound tenderness, or Murphy's sign were detected. Laboratory studies were not available and his vitals were unremarkable.

Vince has been diagnosed with GSID. **GSID**

About Sucrose

Sucrose is commonly known as sugar or table sugar. It consists of one molecule of glucose and one molecule of fructose. Sugars like sucrose that consist of three or more molecules are labeled disaccharides and are joined by a special chemical link called a glycosidic bond that binds carbohydrates together. Glucose and fructose are digested, absorbed and metabolized separately, but both result in the same end product which your body uses for energy.

Digestion

Sucrose digestion does not begin until the sugar reaches the small intestine. The small intestine is lined with finger-like projections called microvilli. The microvilli are known collectively as the brush border. The microvilli absorb nutrients from food as it passes through the small intestine. Your body cannot absorb disaccharides as is, so it must first break sucrose down into its component parts. Through a process called hydrolysis, water assists in breaking the glycosidic bond to separate the glucose and fructose molecules. One molecule of water is needed for each molecule of sucrose. This reaction naturally occurs very slowly. The presence of sucrase, an enzyme in the small intestine, accelerates this reaction.

Absorption

As separate monosaccharides, or one-molecule sugars, glucose and fructose are free for absorption. Both sugars permeate the intestinal lining through distinct complex transporters into the bloodstream and veins, or hepatic portal system. This special system is one of only three in the entire body that does not return blood directly to the heart. Instead, this system carries its blood that contains all absorbed nutrients to the liver for further processing.

Metabolism

After leaving the liver, glucose, the primary source of energy, travels to nearly every cell in the body, and the hormone insulin is assimilated into cells. Glucose is then converted to pyruvate through a process called glycolysis, the metabolic breakdown of glucose. An acidic compound, Pyruvate, then enters either aerobic or anaerobic respiration to produce energy. The metabolism of fructose occurs through fructolysis, a process similar to, but more complex than, glycolysis. Fructolysis results in glucose-like products that can enter respiration for energy production. Unlike glycolysis, which can take place in nearly all tissues, fructolysis occurs primarily in the liver.

Storage

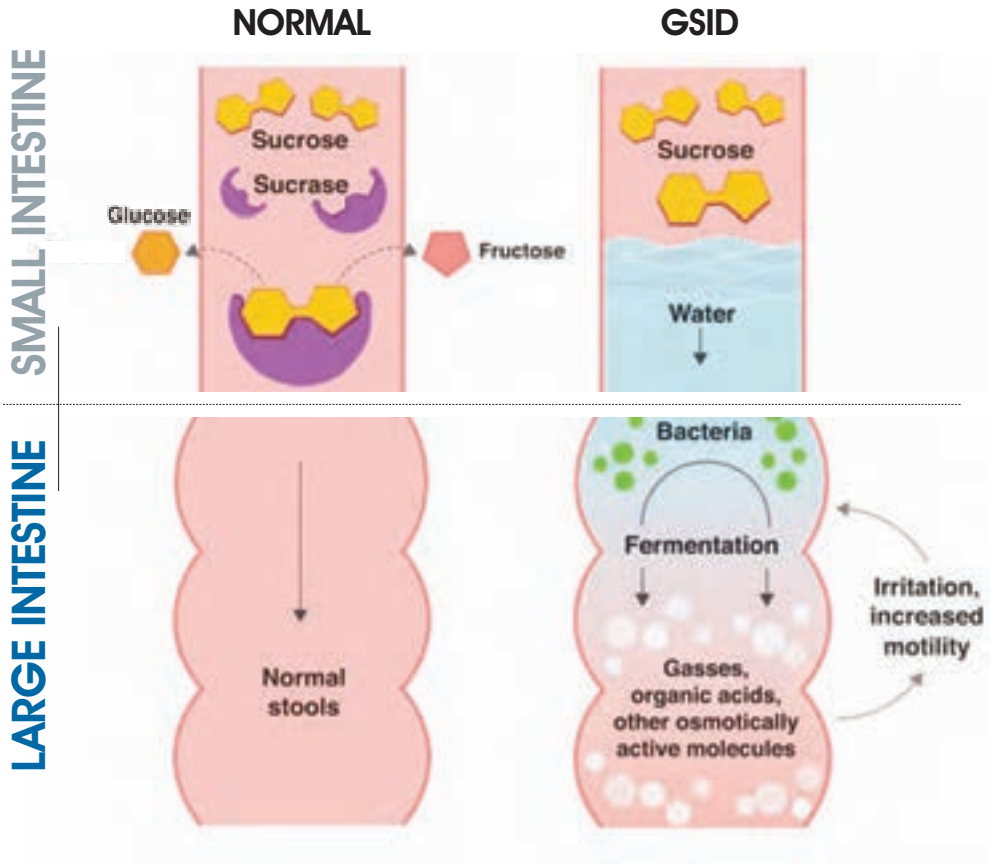
Glucose that is not immediately used for energy undergoes a process called glycogenesis. This process links individual glucose subunits into long chains, known as glycogen, through a chemical bond. Glycogen is then stored in the liver and muscles and hydrolyzed (the break down by chemical reaction with

water) back into glucose as needed, typically between meals and/or while you are sleeping when blood sugar levels are low. When your body reaches its glycogen storage capacity, it converts all remaining glucose into fat, according to "Human Physiology." Fructose is not stored long-term in the body, because the liver metabolizes all fructose to glucose-like molecules.

Complications

A deficiency in the sucrase enzyme, called Genetic Sucrase-Isomaltase Deficiency, can inhibit sucrose digestion and absorption. People with this genetic disorder produce little to no sucrase to hydrolyze (sever) sucrose into glucose and fructose. This allows sucrose to pass in undigested form through the intestines serving as fuel for the naturally occurring bacteria. The bacterial metabolism results in excessive gas, bloating, cramping, abdominal pain, constipation and diarrhea. The lack of glucose absorption decreases energy production and disrupts thousands of daily biochemical processes, which may affect physical growth and development. **GSID**

Sucrose Digestion



GSID Diagnosis

If genetic sucrase-isomaltase deficiency is suspected, there are several approaches that can be implemented to diagnose this deficiency. The first approach is definitive and considered the "gold standard" for the diagnosis of this deficiency.

It is a disaccharidase assay of a small bowel biopsy is obtained through an upper endoscopy (EGD). A sucrose intolerance hydrogen breath test can be administered at home. The results of this test, although not definitive, can give an indication of sucrose maldigestion.

2 Simple Tests

1

Sucrase Enzyme Assay via EGD

2

Sucrose Intolerance Hydrogen Breath Test

Be sure to consult with your healthcare provider to review available options and discuss what is right for you.