RESEARCH
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Relationships among Dietary Intakes and Persistent Gastrointestinal Symptoms in Patients Receiving Enzyme Treatment for Genetic Sucrase-Isomaltase Deficiency

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ABSTRACT

Background Sucrose-isomaltase deficiency (SID) remains underdiagnosed. Absent or reduced enzyme activity promotes diarrhea, abdominal bloating, and flatulence from undigested and malabsorbed disaccharides. Frequency and severity of gastrointestinal symptoms may be associated with the type of carbohydrates consumed.

Objective To characterize the dietary intakes of patients treated with sacrosidase (Sucraid; QOL Medical) for SID and determine relationships between type of carbohydrates, sacrosidase dose, and gastrointestinal symptoms.

Design A prospective 30-day observational study.

Participants/setting Forty-nine patients treated with sacrosidase for ≥3 months were recruited from the enzyme manufacturer's nationwide clinical database between November 2014 and August 2015.

Main outcome measures Dietary energy and nutrient intakes reported during 24-hour diet recall interviews, frequency and severity of gastrointestinal (GI) symptoms, and sacrosidase dose.

Statistical analyses performed Relationships between nutrient intakes, sacrosidase dose, and GI symptoms were evaluated using Spearman ρ correlation coefficients.

Results Sacrosidase dose averaged 5.2±3.1 mL/day. Participants reported 1.3±0.9 bowel movements daily. Having less frequent GI symptoms was associated with higher sacrosidase intake. Energy intakes averaged 1,562.5±411.5 kcal/day in children, 1,964.7±823.6 kcal/day in adolescents, and 1,952.6±546.5 kcal/day in adults. Macronutrient composition averaged 44% carbohydrate, 39% fat, and 17% protein. Average carbohydrate composition was 35% starch, 8% fiber, and 59% sugars. Sucrose and fructose intakes were not associated with GI symptoms. Lactose intake was associated with diarrhea. Maltose intake was associated with nausea, distension, and reflux.

Conclusions Intakes were lower in carbohydrates and higher in fat compared with the Acceptable Macronutrient Distribution Ranges. Sucrose and fructose intakes were not associated with GI symptoms. Higher maltose and lactose intakes were associated with GI symptom frequency and severity. These findings provide evidence to guide nutrition counseling for patients treated for SID.

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SUCRASE-ISOMALTASE DEFICIENCY (SID) IS MOST often an inherited disorder where genetic variants in the sucrase-isomaltase (SI) gene cause protein transport errors resulting in the absence or insufficiency of the disaccharidases sucrase and isomaltase at the brush border of the small intestine lumen. Maltase activity is also reduced because SI accounts for 60% to 80% of its activity in the brush border.[1] Consequently, these maldigested carbohydrates enter the colon, causing excess bacterial fermentation resulting in abdominal distension, cramping, pain, excessive flatulence, and osmotic diarrhea. If untreated, SID leads to inadequate growth and failure to thrive in children as well as weight loss and malnutrition in adults.[2-6]

SID is not typically part of the diagnostic algorithm for managing chronic diarrhea until more common etiologies, such as chronic nonspecific diarrhea (ie, toddler's diarrhea) or irritable bowel syndrome, are explored.[5,7,8] In addition to true congenital SID, an increasing number of patients may have secondary SI deficiency resulting from physical injury of the intestinal brush border, villus atrophy from autoimmune disorders such as celiac disease, or as a result of gastrointestinal infection.[9,10] Medical nutrition therapy using a

low-sucrose/low-starch diet is effective in reducing the frequency and severity of gastrointestinal symptoms. However, the extreme restrictiveness of the diet promotes poor long-term compliance.[5] In combination with a less-severe dietary regimen, an oral sucrase enzyme replacement, sacrosidase (Sucraid; QOL Medical), which hydrolyzes sucrose, has shown efficacy with regard to resolving abdominal cramping, bloating, gas, and watery diarrhea.[4,5,11,12]

Despite these treatment options, some patients treated for SID continue to experience gastrointestinal distress. To our knowledge, no studies have characterized the composition of typical dietary intake in patients treated for SID and whether the intake of specific foods or nutrients contributes to lingering gastrointestinal symptoms once diagnosis and combination treatment have been implemented. The purpose of the present study was to characterize the dietary intakes of children and adults who were being treated with sacrosidase for SID, identify nutrient inadequacies by comparison to the Dietary Reference Intakes, and determine relationships between the amount and types of carbohydrates consumed, sacrosidase dose, and persistent gastrointestinal symptoms.

MATERIALS AND METHODS

Written informed consent was provided and a central institutional review board along with the Vanderbilt University Medical Center Institutional Review Board approved the study protocol. From November 2014 to August 2015, 66 patients were recruited from QOL Medical's nationwide clinical database of patients being treated with sacrosidase and by study advertisement accompanying sacrosidase home shipments. All patients in the database had previously provided a waiver for Health Insurance Portability and Accountability Act authorization for research recruitment. Potential participants were screened by the study team for meeting inclusion criteria that they had been undertaking sacrosidase treatment for ≥3 months, were English speakers, and had a mobile device to enable using the Trial Guide (mProve Health, Arlington, VA) application for providing medication use and gastrointestinal symptom data electronically.

At enrollment, participants were instructed to maintain their current diet and physical activity for the 30-day observational period. Newly prescribed sacrosidase patients typically receive counseling from a registered dietitian nutritionist (RDN) to eliminate sucrose and starch from their diet for 2 weeks while starting sacrosidase therapy and then gradually reintroduce these foods to establish sucrose and starch tolerance. Demographic data (age, sex, race, height, and weight) were self-reported via an online Family Health Questionnaire during the first week of the study (Table 1). A parent or family caregiver provided primary data for participants younger than age 11 years and assisted with data collection for those aged 11 to 17 years.

During the observation period, participants completed three 24-hour diet recall interviews that included obtaining information regarding dietary supplement use. Diet recalls were conducted on 2 weekdays and 1 weekend day and were performed over the telephone by one RDN from the Vanderbilt Diet, Body Composition, and Human Metabolism Core who was trained in using the US Department of Agriculture multipass methodology.[13-15] A food amounts booklet was provided to participants at enrollment to assist with portion size estimation. For safety purposes, participants were instructed that in the case that they noticed a change in gastrointestinal symptom frequency or severity they should telephone the Vanderbilt Diet, Body Composition, and Human Metabolism Core to report their prior 24-hour intake,

Table 1. Characteristics of 49 individuals being treated for sucrase-isomaltase deficiency who participated in an observational study of dietary intake, sacrosidase use, and gastrointestinal symptoms

Characteristic	n	%	Age	BMI[a]	*z* score	Sacrosidase dose (mL/d)[b]
Sex						
Male	32	65.3	—	—	—	—
Female	17	34.7	—	—	—	—
Race/ethnicity						
White, non−Hispanic	44	89.8	—	—	—	—
Other	5	10.2	—	—	—	—
Life stage group[c]						
Age 1-3 y[d]	9	18.4	2.0±0.9	16.2±1.8	−0.07±1.4	4.0±2.1
Age 4-8 y[d]	17	34.6	5.8±1.6	16.0±2.3	−0.85±2.8	6.2±4.0
Age 9-18 y[d]	14	28.6	12.9±1.9	19.2±4.3	−0.31±1.5	6.7±2.9
Age ≥19 y	9	18.4	27.6±10.4	26.0±5.1	—	3.9±2.5

[a]BMI=body mass index.

[b]At the time of this study, Sucraid (QOL Medical) was listed in the Food and Drug Administration's Drug Shortages Database. To avoid a drug shortage, QOL Medical the Food and Drug Administration made Sucraid lots available under a consent process, requiring both the prescribing physician and the patient (or primary caregiver) to sign a consent waiver. Three of 49 participants in this study received one of the consented Sucraid lots while enrolled. None have reported adverse reactions or events.

[c]Life stage group as defined in the Dietary Reference Intakes with age ranges of 1 to 3 years, 4 to 8 years, 9 to 13 years, 14−18 years, and ≥19 years. The 9 to 13 years and 14 to 18 years age groups were collapsed into one adolescent group yielding four life stage categories due to limited sample size.

[d]A parent or family caregiver provided primary data for participants younger than age 11 years and assisted with input for those aged 11 to 17 years.