2017

Sucrase-Isomaltase Deficiency, an Under Considered Diagnosis

Lindsay Venn
University of North Dakota

Follow this and additional works at: https://commons.und.edu/pas-grad-posters

Part of the Digestive System Diseases Commons, and the Nutritional and Metabolic Diseases Commons

Recommended Citation
Venn, Lindsay, "Sucrase-Isomaltase Deficiency, an Under Considered Diagnosis" (2017). Physician Assistant Scholarly Project Posters. 45.
https://commons.und.edu/pas-grad-posters/45

This Poster is brought to you for free and open access by the Department of Physician Studies at UND Scholarly Commons. It has been accepted for inclusion in Physician Assistant Scholarly Project Posters by an authorized administrator of UND Scholarly Commons. For more information, please contact zeinehyousif@library.und.edu.
Sucrase-Isomaltase Deficiency, an Under Considered Diagnosis
Lindsay Venn PA-S
Department of Physician Assistant Studies, University of North Dakota School of Medicine & Health Sciences
Grand Forks, ND 58202-9037

Abstract
• Persistent gastrointestinal (GI) complaints are a common problem in patients presenting to family practice for care.
• Irritable bowel syndrome (IBS) is a diagnosis that adult patients are labeled with when clinicians have exhausted their differential list and no other cause of their GI symptoms can be confirmed.
• Sucrase-isomaltase deficiency (SID) has been historically shown to present early in life as a congenital disorder, but researchers are now recognizing occurrence later in life as a mild or secondary disorder.
• The literature emphasizes the disconnecting prevalence of sucrose-isomaltase deficiency (SID), the persistent symptoms associated with it, and the need for further research, especially in the adult population.
• SID testing is cost effective, very reliable, and completely non-invasive
• It has been proven that after treatment of SID with sucrase replacement enzyme, patients were able to return to normal sucrose digestion and had resolution of their symptoms.
• Educating practitioners about SID and providing options for diagnosis and treatment are the first steps in helping the many patients that suffer from chronic bowel problems.

Introduction
• Patients with an IBS diagnosis account for $1.7-10 billion in medical costs and an additional $10-20 billion of indirect costs in America annually (Cash & Chey, 2004).
• 2.4-3.5 million yearly physician visits are specifically for patients with IBS symptoms (Cash & Chey, 2004).
• The variety of presentations and differing levels of severity of GI symptoms lead to many patients being misdiagnosed with a functional GI disorder (Puntis & Zamvar, 2015).
• The purpose of this scholarly project is to educate practitioners on the symptomology, the ease and low cost of diagnostic testing, and the treatment options for patients with SID.

Literature Review
Pathophysiology of SID:
• Sucrase-isomaltase (SI) is one of the main enzymes responsible for breaking down small starch units into glucose which can then be transported through the mucosal sodium dependent glucose cotransporter gate for utilization in the body (Lin, Hamaker, & Nicholls, 2012).
• When SI is not available or not functioning properly, a variety of adverse symptoms can occur.
• Patients with either SID or IBS complain of diarrhea, abdominal problems, dyspeptic symptoms and worsening of these symptoms following eating (Puntis & Zamvar, 2015; Urich et al., 2012).

Diagnosis of SID:
• Robayo-Torres et al. (2009) evaluated the sensitivity and specificity of 13C-breath tests in patients already confirmed to have CID by duodenal biopsy. Their results were encouraging, they found 100% specificity and 100% sensitivity for determining low sucrose activity by 13C-breath testing when compared to duodenal biopsy.

Treatment options for SID:
• Sucraid is the only FDA approved enzyme replacement available.
• Direct starch digestion by sucrase isomaltase is more efficient than the results are returned directly to the provider so follow up is guaranteed (Sucraid, 2016).

Discussion
• Daileda et al. (2016) say it best, “the diagnosis of functional GI disease is usually made without evaluations of carbohydrate activity, although symptoms from carbohydrate intolerance can overlap”.
• The focus has been directed at a way to more accurately, efficiently, and economically diagnose IBS, rather than testing for SID.
• Symptoms of IBS and SID are similar in many ways, both are chronic and unrelenting and can include diarrhea, abdominal distention, bloating and abdominal pain.
• Nicholas et al. (2012) found that 9.3% of 30,334 patients tested for disaccharidase deficiency were found to be sucrase deficient.
• El-Chamas, Williams and Miranda (2015) found 37% of 203 patients evaluated for chronic abdominal pain were actually sucrase deficient.
• Cohen (2016) cited several studies in his paper where the incidence of SID was found to range from 5-21% in patients, and disaccharidase deficiencies were found in 29.4% of patients presenting with GI symptoms.
• Daileda et al. (2016) found in their meta-analysis that 9% of 34,753 patients evaluated with EGD for possible disaccharidase deficiencies were sucrase deficient.
• 13C-breath testing for SID is free to patients and the results are returned directly to the provider so follow up is guaranteed (Sucraid, 2016).

Applicability to Clinical Practice
• Up to 20% of adults experience unrelenting symptoms of abdominal pain and altered bowel habits and have not been diagnosed with a cause (Eswaran et al., 2011).
• Once SID is confirmed, treatment is quite effective with all patients who were tested demonstrating full correction of SID symptoms with Sucraid supplementation and diet restriction (Robayo-Torres et al., 2009).
• Practitioners can utilize the 13C-breath testing and subsequent Sucraid support and therefore improve the quality of life in 10-20% of their patients with chronic abdominal problems.
• This could lead to a 10-20% reduction in patient and practitioner time as well as medical expense if SID testing was considered earlier.
• All practitioners should add SID to their differential diagnosis list and implement testing for their patients.

Acknowledgements
I would like to give thanks to Professor Metzger and Professor Sieg for all their help in preparing this scholarly project. I would also like to thank my family for all their support and encouragement during this program, it hasn’t been easy on them and they went above and beyond to help make this all possible.

Statement of the Problem
• Many patients suffer needlessly with sucrose-isomaltase deficiency (SID) because clinicians are unfamiliar with and uneducated regarding this disease, its diagnosis, and its treatments.
• More studies are needed to demonstrate the potential prevalence of SID in these misdiagnosed older patients and more clinicians need to be made aware of the possibility of SID as a true condition leading to chronic GI problems.

References