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Sucrase-Isomaltase Deficiency, an Under Considered Diagnosis
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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 disconcerting 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.

Research Questions

• In patients with chronic abdominal pain, flatulence, diarrhea and/or constipation, are providers missing the diagnosis of SID simply because they aren’t testing for it?
• Are clinicians giving the broad diagnosis of IBS to patients who may have SID?
• Should testing for SID be included in preliminary diagnostics for patients with chronic adverse GI symptoms?

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 then can be transported through the mucosal sodium dependent glucose cotransporter gate for utilization in the body (Lin, Hamaker, & Nichols, 2012).
• When SI is not available or not functioning properly, a variety of adverse symptoms can occur.

Comparing SID and IBS:

• IBS is a functional disorder, one without physical or biological etiology, therefore, there aren’t any diagnostic studies that can be done to definitively diagnose it (El-Salhy, 2013).
• Patients with either SID or IBS complain of diarrhea, abdominal pain, dyspeptic symptoms and worsening of these symptoms following eating (Puntis & Zamvar, 2015; Uhrich et al., 2012).

Diagnosis of SID:

• Robayo-Torres et al. (2009) evaluated the sensitivity and specificity of 1C-breath tests in patients already confirmed to have CSID by duodenal biopsy. Their results were encouraging, they found 100% specificity and 100% sensitivity for determining low sucrose activity by 1C-breath testing when comparing to duodenal biopsy.

Treatment options for SID:

• Sucraid is the only FDA approved enzyme replacement available. In the study evaluating 1C-breath testing, the authors compared patient’s test values before and after initiation of Sucraid and found that patients were able to return to normal sucrose digestion with addition of Sucraid (Robayo-Torres et al., 2009).

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 (Eswarzim 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 1C-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 early.
• All practitioners should add SID to their differential diagnosis list and implement testing for their patients.

Discussion

• Daieda et al. (2016) say it best, “the diagnosis of functional GI disease is usually made without evaluations of disaccharide activity, although symptoms from carbohydrate intolerance can overlap”.
• The focus has been directed at a way 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.
• Nichols et al. (2012) found that 9.3% of 30,334 patients tested for disaccharide deficiency were found to be 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 disaccharide deficiencies were found in 29.4% of patients presenting with GI symptoms.
• Daieda et al. (2016) found in their meta-analysis that 9% of 34,753 patients evaluated with EGD for possible disaccharide deficiencies were sucrase deficient.
• 1C-breath testing for SID is free to patients and the results are returned directly to the provider so follow up is guaranteed (Sucraid, 2016).
• The validity of the testing is excellent with 100% sensitivity and specificity (Robayo-Torres et al., 2009).

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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