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CLINICAL CASE

Congenital sucrase–isomaltase deficiency: A case report

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KEYWORDS
Congenital sucrase–isomaltase deficiency;
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Abstract
Background: Congenital sucrase–isomaltase deficiency (CSID) is an autosomal recessive disease characterized by absent sucrase activity with variable decrease in isomaltase activity. The prevalence of CSID in Portuguese population is unknown and there are few reported cases.
Case report: We report the case of a six-month-old male infant admitted for chronic profuse diarrhea and failure to thrive that began after food diversification. The investigation showed that he had CSID. The therapeutic option was the addition of baker's yeast to the diet which was followed by complete resolution of symptoms and excellent weight recovery.
Discussion: This case highlights the relevance of clinical observation and awareness in a condition where diagnosis is essentially clinical. The available therapeutic options are addressed with pragmatic use of baker's yeast.

PALAVRAS-CHAVE
Deficiência congénita de sacarase-isomaltase;
Sacrosidase;
Fermento;
Saccharomyces cerevisiae

Défice congénito de sacarase-isomaltase: relato de um caso

Resumo
Introdução: O défice congénito de sacarase-isomaltase é uma doença autossómica recessiva caraterizada pela ausência da atividade da sacarase e diminuição variável da atividade da isomaltase. A prevalência desta patologia na população portuguesa é desconhecida e há poucos casos divulgados.

Abbreviations: CSID, congenital sucrase–isomaltase deficiency; DNA, deoxyribonucleic acid; IgE, immunoglobulin; EPCR, polymerase chain reaction; SI, sucrase–isomaltase.

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Relato de caso: Lactente masculino de 6 meses, internado para estudo de diarreia crónica profusa coincidente com o início da diversificação alimentar e associada a má evolução ponderal. A investigação mostrou tratar-se de uma deficiência congénita de sacarase-isomaltase. A medida terapêutica escolhida foi a adição de fermento de padeiro na dieta, com resolução da sintomatologia e excelente recuperação ponderal.
Discussão: Discute-se a importância de uma elevada suspeição clínica numa patologia cujo diagnóstico é essencialmente clínico e também as opções terapêuticas disponíveis, com ênfase no uso do fermento de padeiro.

1. Introduction

Congenital sucrase–isomaltase deficiency (OMIM #222900) is caused by homozygous or compound heterozygous mutation in the *SI* gene, which encodes sucrase–isomaltase on chromosome 3q26.

Sucrase–isomaltase is an enterocyte-specific small intestine brush-border membrane disaccharidase. It is required for hydrolysis of sucrose and some starches. Upon ingestion of disaccharides and oligosaccharides, the failure to breakdown sucrose into fructose and glucose results in osmotic-fermentative diarrhea.

CSID is the most common congenital disorder of carbohydrate metabolism. Its estimated prevalence in North America and Europe ranges from 0.05% to 0.2%,[1] although this diagnosis is believed to be frequently missed.

Onset usually occurs during infancy after weaning from breast milk or lactose-only formula onto foods containing sucrose or starch. Clinical manifestations include osmotic-fermentative diarrhea, abdominal distension and discomfort, flatulence, vomiting and diaper rash.[2] Severe symptoms may lead to failure to thrive, dehydration and malnutrition. Adolescents and adults may present with signs of 'irritable bowel syndrome'. Carbohydrates result in a dose-dependent acceleration of colonic transit[3] and therefore symptoms may only occur with the ingestion of large amounts of sucrose.[2]

CSID is a heterogeneous disorder. Identified mutations lead to a range of posttranslational defects resulting in an absence of sucrase activity and varying degrees of isomaltase deficiency. Heterozygotes have intermediate enzyme values and are usually asymptomatic in adulthood, but may have mild symptoms in infancy.

Several tests can be used to diagnose CSID, with measurement of intestinal disaccharides' activities being the gold standard.[2] It will show complete or almost complete lack of sucrase/isomaltase activity with normal lactase activity and normal villous architecture.

The hydrogen breath test is a non-invasive method for detecting carbohydrate malabsorption. This test is based on the fact that unabsorbed sugar is converted to hydrogen gas by colonic bacteria, which is eliminated *via* expired air.[3] However, it is not specific for the diagnosis of CSID and false-negative results may be obtained because of many factors affecting the hydrogen production.[3] An evolution of the hydrogen breath test is the measurement of isotope-labeled CO_2 in breath using ^{13}C or ^{14}C by mass spectrometers. A sucrose breath test for screening and confirmation of CSID using a novel non-invasive ^{13}C-sucrose labeled substrate has been developed and validated, and is an accurate and specific noninvasive confirmatory test for CSID.[4] However, obtaining breath samples may be difficult in small children. Evidence of unabsorbed sugars (Clinitest®) also provides indirect evidence of poor carbohydrate digestion. However, Clinitest® looks for reducing sugars in the stool and sucrose, as stated later, is not in its natural form a reducing sugar. Therefore, negativity of Clinitest is not actually a false negative test; it is a true negative test in the context of Sucrase Isomaltase deficiency when looking for sucrose.

Fecal carbohydrates can also be measured by high-performance liquid chromatography, but this test has some false negatives due to transport times required, that allow consumption of sugars by fecal flora. Other diagnostic methods available include the glycemic curve and stool tests (fecal pH lower than 5.5 with or without reducing sugars without steatorrhea).[3,5] Molecular genetic test for mutation in *SI* gene is also available.

The mainstay of treatment is a lifelong adherence to a sucrose- and starch-restricted diet.[5] Effective enzyme replacement with whole yeast cells has been reported.[6] Purified yeast enzyme, sacrosidase, is a highly effective therapy and allows a normal diet.[4]

CSID's prognosis is good. Affected patients tend to experience spontaneous improvement of their symptoms with age, because colonic bacteria become able to metabolize non absorbed carbohydrates into organic acids (lactic acid and short chain fatty acids), most of which are then absorbed.[3] Patients learn to identify their tolerance limits, but remain exposed to occasional episodes of diarrhea when they ingest amounts of carbohydrates that exceed their ability to metabolize and digest. This can explain the small number of diagnosed cases.

2. Case report

The patient is a 6-months-old Caucasian male infant admitted in our hospital for chronic diarrhea and failure to thrive. He is the only child of young, healthy and non-consanguineous parents. His father had a history of frequent episodes of diarrhea in youth, which were never the subject of any investigation.