Intestinal saccharidase deficiencies

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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references


Reactome database release: 79

This document contains 1 pathway and 5 reactions (see Table of Contents)

https://reactome.org
Intestinal saccharidase deficiencies

Stable identifier: R-HSA-5659898

Diseases: intestinal disaccharidase deficiency

Defects in in two enzymes required for intestinal digestion of dietary carbohydrate, lactase (LCT, a domain of lactase-phlorizin hydrolase protein) and sucrase-isomaltase (SI), are annotated here. The first affects nursing infants; the second affects individuals after weaning.

The disaccharide lactose is a major constituent of human breast milk. To be taken up from the gut in the nursing infant, this sugar must first be hydrolyzed by LCT present on the external face of enterocytes in microvilli of the small intestine. Mutations that disrupt LCT activity are associated with acute illness in newborn children as lactose fermentation by gut bacteria leads to severe diarrhea. The condition is effectively treated by feeding affected infants a lactose-free formula. This congenital disease is distinct from the down-regulation of LCT expression after weaning in many human populations that is associated with a milder form of lactose intolerance in adults (Jarvela et al. 2009).

The starch in a post-weaning diet is digested by amylases to di- and oligosaccharides that must be further digested to monosaccharides in order to be taken up from the lumen of the small intestine into endothelial cells of the intestinal brush border. If they are not digested, a process in which enterocyte-associated SI plays a central role, they remain in the gut lumen and are fermented by gut bacteria, leading to osmotic and fermentative diarrhea (Naim et al. 2012; Van Beers et al. 1995).

Literature references


**Editions**

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Defective SI does not hydrolyze Mal

Location: Intestinal saccharidase deficiencies

Stable identifier: R-HSA-5659922