

Disaccharidase, Tissue

FOR THE DIAGNOSIS OF INHERITED OR ACQUIRED DEFICIENCIES OF THE INTESTINAL DISACCHARIDASES

Clinical Background

Dietary carbohydrates are the major exogenous source of glucose. Nutritionally significant carbohydrates include the disaccharides that consist of two covalently linked sugar molecules. Because the small intestine is normally impermeable to disaccharides, the activity of intestinal disaccharidases is required for absorption of their component monosaccharides. The four enzymes or enzyme complexes for disaccharide hydrolysis (disaccharidases) are lactase-phlorizin hydrolase, sucrase-isomaltase, maltase-glucoamylase, and trehalase. The table below shows the substrate(s) hydrolyzed by each of these enzymes and the monosaccharide products produced.

Enzyme	Substrate(s)	Products
Lactase	Lactose (β -1,4 linkage)	Glucose; Galactose
Sucrase-Isomaltase		
Sucrase	Sucrose (α -1,2 linkage)	Glucose; Fructose
	Maltose (α -1,4 linkage)	Glucose
Isomaltase	Isomaltose (α -1,6 linkage)	Glucose
	Maltose (α -1,4 linkage)	Glucose
Maltase-Glucoamylase	Maltose (α -1,4 linkage)	Glucose
Trehalase	Trehalose (α -1,1 linkage)	Glucose

Disease Overview

- Disaccharidase deficiencies can be congenital (primary), acquired, or secondary.
- Congenital deficiencies are rare.
- Acquired lactase deficiency is the most common form of carbohydrate intolerance.
- Secondary deficiencies occur in conditions that damage the small bowel mucosa, such as celiac sprue, tropical sprue, or acute intestinal infections. Recovery from the underlying disease may restore enzyme activities.

Pathophysiology

Disaccharides are normally hydrolyzed into monosaccharides by disaccharidases located in the brush border of small bowel enterocytes. Decreased or absent activities of one or more of these enzymes in the intestinal mucosa can result in carbohydrate maldigestion. The undigested disaccharides cause an osmotic load that attracts water and electrolytes into the bowel, causing watery diarrhea. Bacterial fermentation of carbohydrates in the colon produces gases resulting in excessive flatulence, bloating and distention, and abdominal pain.

Indications for Ordering

To confirm a deficiency of one or more intestinal disaccharidases.

Additional Ordering Notes

- The collection of two 5 mg biopsies of small bowel is recommended to allow for repeat testing if necessary.
- Place the collected tissue on the wall of a small, tightly capped plastic tube **without any supporting media**. ARUP recommends the use of a 2.0 mL flat-bottom microtube.
- Specimens should be frozen within two hours of collection.

Methodology

- Small bowel mucosa is homogenized in buffer and then incubated with a disaccharide substrate (i.e., lactose, sucrose, maltose, and palatinose) in four separate reactions. Disaccharidases from the mucosa catalyze the hydrolysis of the disaccharides into monosaccharides, including the monosaccharide glucose. The liberated glucose is measured, along with the total protein concentration of the homogenate. The activities of each enzyme are reported in units of $\mu\text{mol}/\text{min}/\text{g}$ protein.
- The lactose reaction determines the activity of lactase. The sucrose reaction determines the activity of the sucrase domain of the sucrase-isomaltase complex. Because maltose is hydrolyzed by maltase, sucrase, and isomaltase, the maltose reaction reflects the activities of each of these enzymes, although ~80 percent of the maltase activity is accounted for by the sucrase-isomaltase complex and ~20 percent by maltase-glucoamylase. The palatinose reaction determines the α -1,6 glycosidic hydrolysis activity of isomaltase.

Interpretation

Low disaccharidases activities with normal histology suggest a congenital or acquired deficiency. The presence of abnormal histology suggests a secondary deficiency.

Limitations

Trehalase activity is not detected by this test.

References

- Dalqvist A. Method for the assay of intestinal disaccharidases. *Analytical Biochemistry* 1964;7:18–25.
- Robayo-Torres CC, Quezada-Cavillo R, Nichols BL. Disaccharide digestion: clinical and molecular aspects. *Clinical Genomics* 2006;4:276–87.
- Hertzler SR, et al. Intestinal disaccharidase depletions. In *Modern nutrition in health and disease*, 10th ed. M Shils, et al, eds. 2006; Philadelphia: Lippincott Williams & Wilkins.

Test Information

2002247 **Disaccharidase, Tissue**

For specific collection, transport, and testing information, refer to the ARUP Web site at www.aruplab.com.

For information on test selection, ordering, and interpretation, refer to ARUP Consult® at www.arupconsult.com.