

Overview

Useful For

Evaluation of patients who present with signs or symptoms suggestive of disaccharidase disorders

This test is **not intended for** carrier detection.

Genetics Test Information

Diagnostic testing for patients with clinical signs and symptoms suspicious of a disaccharidase deficiency.

Method Name

Spectrophotometry

NY State Available

Yes

Specimen

Specimen Type

Tissue

Shipping Instructions

Freeze immediately after collection and ship frozen.

Specimen Required

Specimen Type: Tissue

Source: Intestinal biopsy

Container/Tube: Clean, screw-topped plastic vial

Specimen Volume: 5 mg

Collection Instructions: Specimen should **not** be placed on gauze or filter paper, nor should any saline, support or embedding material be added.

Forms

[Biochemical Genetics Patient Information \(T602\)](#)

Reject Due To

Specimens sent in formaldehyde, saline, or any supportive media Reject
 Gauze
 Filter paper
 Foil
 Swabs

Specimen Minimum Volume

See Specimen Required

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Tissue	Frozen (preferred)	28 days	OTHER

Clinical & Interpretive

Clinical Information

Disaccharidases in the small intestines are responsible for the breakdown of disaccharides (double sugars) into monosaccharides (simple sugars). Patients with a deficiency of 1 or more disaccharidase enzymes can experience intolerance to foods containing complex sugars resulting in a range of gastrointestinal symptoms including diarrhea or constipation, abdominal pain and cramping, gas, bloating, and nausea. In addition, patients may experience malnutrition, weight loss, or failure to thrive. Given the nonspecificity and frequency of abdominal symptoms, misdiagnosis or a diagnostic delay of disaccharide deficiencies may occur.

Primary and secondary causes of disaccharidase deficiencies exist, and age of onset may vary from birth through adulthood. Primary causes are rare and result from genetic alterations in a variety of genes. Secondary deficiencies typically result from small intestinal mucosal damage. Treatment of both primary and secondary disaccharidase deficiencies involves dietary management. While primary deficiencies require lifelong treatment, secondary disaccharidase deficiencies may require treatment only until the intestinal lining recovers.

Reference Values

Lactase: > or =14.0 nmol/min/mg protein

Sucrase: > or =19.0 nmol/min/mg protein

Maltase: > or =70.0 nmol/min/mg protein

Palatinase: > or =6.0 nmol/min/mg protein

Glucoamylase: > or =8.0 nmol/min/mg protein

Interpretation

Quantitative values of lactase, sucrase, maltase, palatinase, and glucoamylase are reported. Clinical interpretation of results is provided.

Cautions

This test will not distinguish between primary and secondary causes of disaccharidase deficiencies.

Clinical Reference

1. Cohen SA, Oloyede H, Gold BD, Mohammed A, Elser HE: Clinical characteristics of disaccharidase deficiencies among children undergoing upper endoscopy. *J Pediatr Gastroenterol Nutr.* 2018 Jun;66 Suppl 3:S56-S60
2. Semenza G, Auricchio S, Mantei N. : Small-intestinal disaccharidases. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. *The Online Metabolic and Molecular Bases of Inherited Disease.* McGraw Hill; 2019. Accessed August 23, 2021. Available at <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225081608>

Performance**Method Description**

Disaccharidase activities are measured by incubating intestinal homogenates with an appropriate disaccharide substrate, which releases glucose. Glucose production is determined by a glucose oxidase reagent mixture that oxidizes glucose to produce peroxide, which reacts with phenol aminophenazone to form a red quinoneimine dye. The concentration of quinoneimine is determined spectrophotometrically and the concentration of liberated glucose is calculated from a glucose standard curve. The amount of glucose produced is proportional to the disaccharidase activity in the intestinal homogenate. Disaccharidase activities are expressed as nanomoles of glucose per minute per milligram of protein (nmol/min/mg prot).(Unpublished Mayo method)

PDF Report

No

Specimen Retention Time

Biopsy homogenate: 1 month

Performing Laboratory Location

Rochester

Fees & Codes**Test Classification**

This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82657

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
DSAC	Disaccharidase Activity Panel, Ts	57765-0

Result ID	Reporting Name	LOINC®
608237	Lactase	81315-4
608238	Sucrase	30372-7
608239	Maltase	30315-6
608240	Palatinase	81314-7
608241	Glucoamylase	81322-0
608242	Interpretation	57779-1
608243	Reviewed By	18771-6