

Test Definition: BTDZ

Biotinidase Deficiency, BTD Full Gene Analysis,
Varies

Reporting Title: BTD Gene, Full Gene Analysis

Performing Location: Rochester

Shipping Instructions:

Specimen preferred to arrive within 96 hours of draw.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call

800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions:

1. Invert several times to mix blood.

2. Send specimen in original tube.

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:

- -Informed Consent for Genetic Testing (T576)
- -Informed Consent for Genetic Testing-Spanish (T826)
- 2. Molecular Genetics: Biochemical Disorders Patient Information (T527) in Special Instructions

Specimen Type	Temperature	Time	Special Container	
Varies	Ambient (preferred)			
	Frozen			
	Refrigerated			

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
53463	Result Summary	Alphanumeric		50397-9
53464	Result	Alphanumeric		82939-0
53465	Interpretation	Alphanumeric		69047-9
53466	Additional Information	Alphanumeric		48767-8
53467	Specimen	Alphanumeric		31208-2
53468	Source	Alphanumeric		31208-2
53469	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.



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No

CPT Code Information:

81404-BTD (biotinidase) (eg, biotinidase deficiency), full gene sequence

Reference Values:

An interpretive report will be provided.