
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.

Specimen Minimum Volume:

0.5 mL

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	Type	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.

Supplemental Report:

No

CPT Code Information:

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

Reference Values:

An interpretive report will be provided.