
Reporting Title: BTK Gene, Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

Targeted testing for familial variants (also called site-specific or known variants testing) is available for variants identified in the BTK gene. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about testing option, call 800-533-1710.

Additional Testing Requirements:

To confirm a diagnosis of X-linked agammaglobulinemia in male patients, the preferred approach is to order this test concurrently with BTK / Bruton Tyrosine Kinase, Protein Expression, Flow Cytometry, Blood.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

Bruton Tyrosine Kinase (BTK) Gene Sequencing Patient Information form (T620) is highly recommended. Testing may proceed without the patient information. However, it aids in providing a more thorough interpretation. Ordering providers are strongly encouraged to complete the form and send it with the specimen.

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.

Submit only 1 of the following specimens:

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 whole blood specimen in original tube. Do not aliquot.
- Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB /Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB /Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Minimum Volume:

Blood: 1 mL; Skin biopsy or cultured fibroblasts: See Specimen Required

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:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing (Spanish) (T826)

2. Bruton Tyrosine Kinase (BTK) Gene Sequencing Patient Information form (T620)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619761	Test Description	Alphanumeric		62364-5
619762	Specimen	Alphanumeric		31208-2

Result ID	Reporting Name	Type	Unit	LOINC®
619763	Source	Alphanumeric		31208-2
619764	Result Summary	Alphanumeric		50397-9
619765	Result	Alphanumeric		82939-0
619766	Interpretation	Alphanumeric		69047-9
619767	Additional Results	Alphanumeric		82939-0
619768	Resources	Alphanumeric		99622-3
619769	Additional Information	Alphanumeric		48767-8
619770	Method	Alphanumeric		85069-3
619771	Genes Analyzed	Alphanumeric		82939-0
619772	Disclaimer	Alphanumeric		62364-5
619773	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

81406
88233- Tissue culture, skin, solid tissue biopsy (if appropriate)
88240- Cryopreservation (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.