
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.

- 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		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
619761	Test Description	Alphanumeric		62364-5
619762	Specimen	Alphanumeric		31208-2
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	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes

Result Codes for Reflex Tests:

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