



Reporting Title: UBA1 Mutation, Quant, ddPCR, V

Performing Location: Rochester

Ordering Guidance:

This test is intended for patients with clinical symptoms and other pertinent laboratory findings raising concern for VEXAS syndrome. These may include, but are not limited to, systemic or localized (eg, ear, orbital, skin) tissue inflammation presenting as rheumatologic disease, abnormal (usually low) whole blood counts, macrocytic anemia, characteristic microscopic changes in the bone marrow, as well as others.

Shipping Instructions:

1. Both refrigerated and ambient specimens must arrive within 7 days of collection.
2. Collect and package specimen as close to shipping time as possible.

Necessary Information:

The following information is required:

1. Pertinent clinical history
2. Date of collection
3. Specimen source (blood or bone marrow)

Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD-B) or green top (heparin)

Specimen Volume: 4 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
3. Label specimen as blood.

Specimen Stability: Refrigerated 7 days/Ambient 7 days

Specimen Type: Bone marrow aspirate

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD-B) or green top (heparin)

Specimen Volume: 2 mL

Collection Instructions:

1. Invert several times to mix bone marrow.
2. Send bone marrow specimen in original tube. Do not aliquot.
3. Label specimen as bone marrow.

Specimen Stability: Refrigerated 7 days/Ambient 7 days



Specimen Type: Extracted DNA from blood or bone marrow

Container/Tube: 1.5- to 2-mL tube with indication of volume and concentration of DNA

Specimen Volume: Entire specimen

Collection Instructions:

1. Label specimen as extracted DNA and source of specimen

2. Indicate volume and concentration of DNA on label. The required volume of DNA is at least 50 mcL at a concentration of 50 ng/mcL

Specimen Stability: Frozen (preferred)/Refrigerated

Specimen Minimum Volume:

Whole blood: 4 mL

Bone marrow: 2 mL

Extracted DNA: 50 mcL at 50 ng/mcL

Forms:

1. Hematopathology Patient Information (T676)

2. If not ordering electronically, complete, print, and send a Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)	7 days	
	Ambient	7 days	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
UBA1Q	MP086	Specimen Type: <ul style="list-style-type: none">• Peripheral blood• Bone marrow• Extracted DNA from peripheral blood• Extracted DNA from bone marrow	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
MP086	Specimen Type	Alphanumeric		31208-2
620938	Interpretation	Alphanumeric		59465-5



Result ID	Reporting Name	Type	Unit	LOINC®
620939	Signing Pathologist	Alphanumeric		19139-5

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81479

Reference Values:

An interpretive report will be provided.