



PTPN11 GENE, KNOWN MUTATION, BLOOD
Test ID: PT1K

PROFILE INFORMATION:

Test ID	Reporting Name	Available Separately	Always Performed
PT1KS	PTPN11 Gene, Known Mutation	No	Yes
PT1KQ	PTPN11, Known Mutation Sequencing	No	Yes

ADDITIONAL TESTS:

Test ID	Reporting Name	Available Separately	Always Performed
EXCVM	DNA Extraction, NPL	No	Yes

TESTING ALGORITHM: When this test is ordered, DNA extraction will always be performed at an additional charge.

USEFUL FOR: Genetic testing of individuals at risk for a known *PTPN11* variant

METHODOLOGY: Polymerase Chain Reaction (PCR) followed by DNA sequence analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:

Collection Container/Tube: Lavender top (EDTA)

Specimen Volume: 3 mL

Collection Instructions: Send specimen in original tube.

Additional Information:

1. Include physician name and phone number with the specimen.
2. Transfusions will interfere with testing for up to 4 to 6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte-reduced. Wait 4 to 6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.

FORMS: Noonan Spectrum Gene Testing Patient Information Sheet (**Supply T689**) in Special Instructions section of test catalog for this test (<http://www.mayomedicallaboratories.com>).

NOTE: New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions section of the test catalog for this test.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood EDTA	Ambient (preferred)	
	Refrigerated (acceptable)	

CAUTIONS: This test is for individuals who are at risk for a *PTPN11* variant that has been previously identified in the family. If the familial variant is not known, the familial proband should be screened for a *PTPN11* mutation using the full gene sequence assay (PT11: *PTPN11*, Full Gene Sequence, Blood).

LIST FEE: \$574.00 plus \$165.50 (DNA extraction) = \$739.50 (Total List Fee)

CPT CODE:

PTPN11 Gene, Known Mutation, Blood

83892-Enzymatic digestion

83894-Separation by gel electrophoresis

83898-Amplification, target, each nucleic acid sequence

83912-Interpretation and report

PTPN11, Known Mutation Sequencing

83909 x 2

DNA Extraction, AutoPure

83891

DAY(S) SET UP: 7 days

ANALYTIC TIME: Varies

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Shirley Pokorski, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710