

Reporting Title: Phenylalanine Disorders Gene Panel

Performing Location Rochester

Ordering Guidance:

The recommended first-tier test for disorders of phenylalanine metabolism is quantitative plasma amino acids (AAQP / Amino Acids, Quantitative, Plasma), as well as neurotransmitters in cerebrospinal fluid and pterin metabolite analysis in blood and urine.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

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 Stability Information: Ambient (preferred)/Refrigerated

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	Varies (preferred)	0 hours	

Supplemental Report:

No

CPT Code Information:

81405

81406 x 2

81479

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Available Separately	Always Performed
FIBR	Fibroblast		88233-Fibroblast	No	Yes

	Culture		culture88240-Cry opreservation for biochemical studies		
CRYOB	Cryopreserve for Biochem Studies		88240	No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC
CRYOB	88832	Cryopreserve for Biochem Studies	Alphanumeric		No LOINC Needed
FIBR	8482	Fibroblast Culture	Alphanumeric		96300-9

Reference Values:

An interpretive report will be provided.