Test Definition: MHDKM

Methylmalonic Aciduria and Homocystinuria, cblD Type, Known Mutation

Reporting Title: MMADHC Gene, Known Mutation
Performing Location: Rochester

Specimen Requirements:

This test can only be performed if a mutation has previously been identified in a family member of this individual.

Forms:
1. 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.
2. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:
Specimen Type: 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/Frozen

Specimen Type: Cultured fibroblasts
Container/Tube: T-75 or T-25 flask
Specimen Volume: 1 full T-75 or 2 full T-25 flasks
Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy
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. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).
Specimen Volume: 4-mm punch
Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:
Specimen Type: Blood spot
Container/Tube: Whatman Protein Saver 903 Paper
Specimen Volume: 5 blood spots
Collection Instructions:
1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
2. Do not expose specimen to heat or direct sunlight.
3. Do not stack wet specimens.
4. Keep specimen dry.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. Prenatal specimens can be sent Monday through Thursday and must be received by 5 p.m. CST on Friday in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on the maternal specimen.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type	Temperature	Time
Varies	Varies	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
FBC	CG030	Source: • Autopsy • Chorionic Villi • Products of Conception or Stillbirth	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No
AFC	CG132	Reason For Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
33371	Specimen	Alphanumeric		N/A
33372	Specimen ID	Alphanumeric		N/A

Result ID	Reporting Name	Type	Unit	LOINC®
33373	Source	Alphanumeric		N/A
33374	Order Date	Alphanumeric		N/A
33375	Reason For Referral	Alphanumeric		N/A
33376	Method	Alphanumeric		N/A
33377	Result	Alphanumeric		N/A
33378	Interpretation	Alphanumeric		N/A
33379	Amendment	Alphanumeric		N/A
33380	Reviewed By	Alphanumeric		N/A
33381	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81479

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes
AFC	Amniotic Fluid Culture/Genetic Test		Profile	No	Yes
MCC	Maternal Cell Contamination, B	1	81265	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		31208-2
FBC	16337	Specimen ID	Alphanumeric		N/A
FBC	CG030	Source	Alphanumeric		N/A
FBC	16168	Order Date	Alphanumeric		N/A
FBC	CG033	Reason For Referral	Alphanumeric		42349-1
FBC	16172	Method	Alphanumeric		In Process
FBC	16175	Interpretation	Alphanumeric		69965-2
FBC	16176	Amendment	Alphanumeric		In Process

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FBC	16177	Consultant	Alphanumeric		N/A
FBC	16178	Report Date	Alphanumeric		N/A
AFC	16179	Specimen	Alphanumeric		31208-2
AFC	16338	Specimen ID	Alphanumeric		N/A
AFC	16793	Source	Alphanumeric		N/A
AFC	16180	Order Date	Alphanumeric		N/A
AFC	CG132	Reason For Referral	Alphanumeric		42349-1
AFC	16184	Method	Alphanumeric		In Process
AFC	16187	Interpretation	Alphanumeric		69965-2
AFC	16188	Amendment	Alphanumeric		In Process
AFC	16189	Consultant	Alphanumeric		N/A
AFC	16190	Report Date	Alphanumeric		N/A
MCC	23332	Specimen	Alphanumeric		31208-2
MCC	23333	Specimen ID	Alphanumeric		N/A
MCC	23334	Source	Alphanumeric		N/A
MCC	23335	Order Date	Alphanumeric		N/A
MCC	23338	Result	Alphanumeric		40704-9
MCC	23337	Amendment	Alphanumeric		N/A
MCC	23341	Reviewed By:	Alphanumeric		In Process
MCC	23339	Release Date	Alphanumeric		N/A

Reference Values:

An interpretive report will be provided.