Reporting Title: MMADHC Gene, Full Gene Analysis
Performing Location: Rochester

Specimen Requirements:

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

Specimen Type: Blood spot
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: Ahlstrom 226 filter paper, or Supplemental Newborn Screening Card (Supply T493)
Specimen Volume: 2 to 5 Blood Spots on collection card (Whatman Protein Saver 903 Paper; Ahlstrom 226 filter paper; or Supplemental Newborn Screening Card, Supply T493)
Collection Instructions:
1. An alternative blood collection option for a patient >1 year of age is finger stick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry
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

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
33360	Specimen	Alphanumeric		N/A
33361	Specimen ID	Alphanumeric		N/A
33362	Source	Alphanumeric		31208-2
33363	Order Date	Alphanumeric		N/A
33364	Reason For Referral	Alphanumeric		42349-1
33365	Method	Alphanumeric		49549-9
33366	Result	Alphanumeric		N/A
33367	Interpretation	Alphanumeric		N/A
33368	Amendment	Alphanumeric		In Process
33369	Reviewed By	Alphanumeric		N/A
33370	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

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		N/A
FBC	16337	Specimen ID	Alphanumeric		N/A
FBC	CG030	Source	Alphanumeric		31208-2
FBC	16168	Order Date	Alphanumeric		N/A
FBC	CG033	Reason For Referral	Alphanumeric		42349-1
FBC	16172	Method	Alphanumeric		49549-9
FBC	16175	Interpretation	Alphanumeric		69965-2
FBC	16176	Amendment	Alphanumeric		In Process
FBC	16177	Consultant	Alphanumeric		N/A
FBC	16178	Report Date	Alphanumeric		N/A

Reference Values:

An interpretive report will be provided.