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

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

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		
33361	Specimen ID	Alphanumeric		
33362	Source	Alphanumeric		
33363	Order Date	Alphanumeric		
33364	Reason For Referral	Alphanumeric		
33365	Method	Alphanumeric		
33366	Result	Alphanumeric		
33367	Interpretation	Alphanumeric		
33368	Amendment	Alphanumeric		
33369	Reviewed By	Alphanumeric		
33370	Release Date	Alphanumeric		

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	MOLECULE ISOLATE NUCLEIC	1	83891		
Billing only	MOLECULE NUCLEIC AMPLI EACH	8	83898		

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	MOLECULE NUCLEIC AMPLI EACH	16	83909		
Billing only	GENETIC EXAMINATION	1	83912		

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		
FBC	16337	Specimen ID	Alphanumeric		
FBC	CG030	Source	Alphanumeric		
FBC	16168	Order Date	Alphanumeric		
FBC	CG033	Reason For Referral	Alphanumeric		
FBC	16172	Method	Alphanumeric		
FBC	16175	Interpretation	Alphanumeric		
FBC	16176	Amendment	Alphanumeric		
FBC	16177	Consultant	Alphanumeric		
FBC	16178	Report Date	Alphanumeric		

Reference Values:

An interpretive report will be provided.