
Reporting Title: FTCD Gene, Known Mutation

Performing Location: Rochester

Specimen Requirements:

Forms:

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

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:

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

Specimen Type: Cultured fibroblasts

Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 full T-75 flask 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®
33536	Specimen	Alphanumeric		N/A
33537	Specimen ID	Alphanumeric		N/A
33538	Source	Alphanumeric		N/A
33539	Order Date	Alphanumeric		N/A
33540	Reason For Referral	Alphanumeric		N/A
33541	Method	Alphanumeric		N/A
33542	Result	Alphanumeric		N/A
33543	Interpretation	Alphanumeric		N/A
33544	Amendment	Alphanumeric		N/A
33545	Reviewed By	Alphanumeric		N/A
33546	Release Date	Alphanumeric		N/A

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	2	83898		

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	NUCLEIC ACID HIGH RESOLUTE	4	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		31208-2
FBC	16337	Specimen ID	Alphanumeric		N/A
FBC	CG030	Source	Alphanumeric		N/A
FBC	16168	Order Date	Alphanumeric		In Process
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
FBC	16177	Consultant	Alphanumeric		N/A
FBC	16178	Report Date	Alphanumeric		In Process

Reference Values:

An interpretive report will be provided.