
Reporting Title: Familial Dysautonomia, Mutation

Performing Location: Rochester

Specimen Requirements:

Forms:

1. Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) 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:

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/Frozen

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 pm 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 both the prenatal and the maternal specimens.

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®
23025	Specimen	Alphanumeric		
23026	Specimen ID	Alphanumeric		
23027	Source	Alphanumeric		
23028	Order Date	Alphanumeric		
23029	Reason For Referral	Alphanumeric		
23030	Method	Alphanumeric		
23031	Results	Alphanumeric		
23032	Interpretation	Alphanumeric		
23033	Amendment	Alphanumeric		
23034	Reviewed By:	Alphanumeric		
23035	Release Date	Alphanumeric		

CPT Code Information:

81260-IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (eg, familial dysautonomia) gene analysis, common variants (eg, 2507+6T->C, R696P)

Amniotic Fluid Culture for Genetic Testing

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

Fibroblast Culture for Genetic Testing
 88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
 88240-Cryopreservation (if appropriate)

 Maternal Cell Contamination, B
 83900 x 6-Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
 83909 x 2-Separation and identification by high-resolution technique (if appropriate)
 83912-Interpretation and report (if appropriate)

 For nonparticipating payers:
 Familial Dysautonomia, Mutation Analysis, IVS20(+6T->C) and R696P
 83890-Molecular isolation or extraction
 83892-Enzymatic digestion
 83896 x 2-Nucleic acid probe, each
 83900-Amplification, target, multiplex, first 2 nucleic acid sequences
 83909-Separation and identification by high-resolution technique
 83912-Interpretation and report
 83914 x 2-Mutation identification by enzymatic ligation or primer extension, single segment, each segment

 Amniotic Fluid Culture for Genetic Testing
 88235-Tissue culture for amniotic fluid (if appropriate)
 88240-Cryopreservation (if appropriate)

 Fibroblast Culture for Genetic Testing
 88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
 88240-Cryopreservation (if appropriate)

 Maternal Cell Contamination, B
 83900 x 6-Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
 83909 x 2-Separation and identification by high-resolution technique (if appropriate)
 83912-Interpretation and report (if appropriate)

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		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		
AFC	16179	Specimen	Alphanumeric		
AFC	16338	Specimen ID	Alphanumeric		
AFC	16793	Source	Alphanumeric		
AFC	16180	Order Date	Alphanumeric		
AFC	CG132	Reason For Referral	Alphanumeric		
AFC	16184	Method	Alphanumeric		
AFC	16187	Interpretation	Alphanumeric		
AFC	16188	Amendment	Alphanumeric		
AFC	16189	Consultant	Alphanumeric		
AFC	16190	Report Date	Alphanumeric		
MCC	23332	Specimen	Alphanumeric		
MCC	23333	Specimen ID	Alphanumeric		
MCC	23334	Source	Alphanumeric		
MCC	23335	Order Date	Alphanumeric		
MCC	23338	Result	Alphanumeric		
MCC	23337	Amendment	Alphanumeric		
MCC	23341	Reviewed By:	Alphanumeric		
MCC	23339	Release Date	Alphanumeric		

Reference Values:

An interpretive report will be provided.