Test Definition: FANCA

Fanconi Anemia C Mutation Analysis, IVS4(+4)A->T and 322delG

Reporting Title: Fanconi Anemia, Mutation Analysis
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: Yellow top (ACD) or lavender top (EDTA)
Acceptable: Any anticoagulant
Specimen Volume: 2.6 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 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®
22970	Specimen	Alphanumeric		31208-2
22971	Specimen ID	Alphanumeric		N/A
22972	Source	Alphanumeric		N/A
22973	Order Date	Alphanumeric		N/A
22974	Reason For Referral	Alphanumeric		42349-1
22975	Method	Alphanumeric		In Process
22976	Result	Alphanumeric		32639-7
22977	Interpretation	Alphanumeric		69047-9
22978	Amendment	Alphanumeric		In Process
22979	Reviewed By:	Alphanumeric		N/A
22980	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81242

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
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

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
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.