Reporting Title: SEPT9 Gene, Known Mutation
Performing Location: Rochester

Specimen Requirements:

Specimen must arrive within 96 hours of collection.

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.

This test can only be performed if a mutation has previously been identified in a family member of this individual.

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 Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
33744	Specimen	Alphanumeric		N/A
33745	Specimen ID	Alphanumeric		N/A
33746	Source	Alphanumeric		N/A
33747	Order Date	Alphanumeric		N/A
33748	Reason for Referral	Alphanumeric		N/A
33749	Method	Alphanumeric		N/A
33750	Result	Alphanumeric		N/A

Result ID	Reporting Name	Type	Unit	LOINC®
33751	Interpretation	Alphanumeric		N/A
33752	Amendment	Alphanumeric		N/A
33753	Review By:	Alphanumeric		N/A
33754	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	GENETIC EXAMINATION	1	83912		

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FUSEQ	DNA Sequence, Follow-up Analysis		Profile	No	No
SEPLD	SEPT9 Gene, Large Del/Dup		Profile	No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FUSEQ	17231	Specimen	Alphanumeric		31208-2
FUSEQ	17232	Specimen ID	Alphanumeric		In Process
FUSEQ	17233	Source	Alphanumeric		In Process
FUSEQ	17234	Order Date	Alphanumeric		In Process
FUSEQ	17235	Result	Alphanumeric		In Process
FUSEQ	17236	Amendment	Alphanumeric		In Process
FUSEQ	17237	Reviewd By	Alphanumeric		In Process
FUSEQ	17238	Released Date	Alphanumeric		In Process
SEPLD	33755	Specimen	Alphanumeric		N/A

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
SEPLD	33756	Specimen ID	Alphanumeric		N/A
SEPLD	33757	Source	Alphanumeric		N/A
SEPLD	33758	Order Date	Alphanumeric		N/A
SEPLD	33759	Result	Alphanumeric		N/A
SEPLD	33760	Amendment	Alphanumeric		N/A
SEPLD	33761	Review By:	Alphanumeric		N/A
SEPLD	33762	Release Date	Alphanumeric		N/A

Reference Values:

An interpretive report will be provided.