**Reporting Title:** ARSA Gene, Known Mutation
**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

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 both the prenatal and maternal specimens.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient OK

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 OK

| Specimen Type | Temperature | Time |
|---|---|---|
| Varies | Varies | |

## Ask at Order Entry (AOE) Questions:

| Test ID | Question ID | Description | Type | Reportable |
|---|---|---|---|---|
| FBC | CG030 | Source:<br>• Autopsy<br>• Chorionic Villi<br>• 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® |
|---|---|---|---|---|
| 33327 | Specimen | Alphanumeric | | |
| 33328 | Specimen ID | Alphanumeric | | |
| 33329 | Source | Alphanumeric | | |

| Result ID | Reporting Name | Type | Unit | LOINC® |
|---|---|---|---|---|
| 33330 | Order Date | Alphanumeric | | |
| 33331 | Reason For Referral | Alphanumeric | | |
| 33332 | Method | Alphanumeric | | |
| 33333 | Result | Alphanumeric | | |
| 33334 | Interpretation | Alphanumeric | | |
| 33335 | Amendment | Alphanumeric | | |
| 33336 | Reviewed By | Alphanumeric | | |
| 33337 | 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 | 2 | 83898 | | |
| 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 |
| 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 | | |

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC® |
|---|---|---|---|---|---|
| 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.