

Bloom Syndrome, Mutation Analysis, 2281del6/ins7

Reporting Title: Bloom Syndrome, Mutation Analysis

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-Congenital Inherited Diseases Patient Information Sheet (Supply T521).

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:

Invert several times to mix blood.
 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

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

Ask at Order Entry (AOE) Questions:

| Test ID | Question ID | Description | Туре | 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 | Туре | Unit | LOINC® |
|-----------|---------------------|--------------|------|--------|
| 22981 | Specimen | Alphanumeric | | |
| 22982 | Specimen ID | Alphanumeric | | |
| 22983 | Source | Alphanumeric | | |
| 22984 | Order Date | Alphanumeric | | |
| 22985 | Reason For Referral | Alphanumeric | | |
| 22986 | Method | Alphanumeric | | |
| 22987 | Result | Alphanumeric | | |
| 22988 | Interpretation | Alphanumeric | | |
| 22989 | Amendment | Alphanumeric | | |
| 22990 | Review By: | Alphanumeric | | _ |
| 22991 | Release Date | Alphanumeric | | |

CPT Code Information:

81209-BLM (Bloom syndrome, RecQ helicase-like) (eg, Bloom syndrome) gene analysis, 2281 del6ins7 variant

Amniotic Fluid Culture for Genetic Testing

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

Fibroblast Culture for Genetic Testing



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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:

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83890-Molecular isolation or extraction

83892-Enzymatic digestion

83896-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-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 |
| мсс | Maternal Cell Contamination, B | | Profile | No | Yes |

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Result Codes for Reflex Tests:

| Test ID | Result ID | Reporting Name | Туре | 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 | | |
| мсс | 23334 | Source | Alphanumeric | | |
| мсс | 23335 | Order Date | Alphanumeric | | |
| мсс | 23338 | Result | Alphanumeric | | |
| мсс | 23337 | Amendment | Alphanumeric | | |
| MCC | 23341 | Reviewed By: | Alphanumeric | | |
| мсс | 23339 | Release Date | Alphanumeric | | |



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Reference Values:

An interpretive report will be provided.