

Reporting Title: BWS/RSS Molecular Analysis**Performing Location:** Rochester**Additional Testing Requirements:**

All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen as this must be a different order number than the prenatal specimen.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

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 whole blood specimen in original tube. Do not aliquot.

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

Additional information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

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.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Prenatal Specimens

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional information:

1. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur.

2. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Acceptable:

Specimen Type: Confluent cultured amniocytes

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured amniocytes from another laboratory.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information: All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Minimum Volume:

Blood: 1 mL; Amniotic Fluid: 10 mL

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULAF	CG767	Reason for Referral	Plain Text	No
CULFB	CG770	Reason for Referral	Plain Text	No

Test ID	Question ID	Description	Type	Reportable
CULFB	CG899	Specimen	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
52845	Result Summary	Alphanumeric		50397-9
52846	Result	Alphanumeric		82939-0
52847	Interpretation	Alphanumeric		69047-9
52848	Reason for Referral	Alphanumeric		42349-1
52849	Specimen	Alphanumeric		31208-2
52850	Source	Alphanumeric		31208-2
52851	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81401-H19 (imprinted maternally expressed transcript [non-protein coding]) (eg, Beckwith-Wiedemann syndrome), methylation analysis

81401-KCNQ1OT1 (KCNQ1 overlapping transcript 1 [non-protein coding]) (eg, Beckwith-Wiedemann syndrome) methylation analysis

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

88235-Tissue culture for amniotic fluid (if appropriate)

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
CULFB	Fibroblast Culture for Genetic Test			No	Yes
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.