
Reporting Title: SMA Diagnostic by Del/Dup**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.

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:**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.

Additional Information: To ensure a minimum DNA amount and concentration, the preferred blood volume must be submitted. Testing may be canceled if the specimen supplied is inadequate.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 14 days

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.

Submit only 1 of the following specimens:**Specimen Type:** Amniotic fluid**Container/Tube:****Preferred:** Screw-capped, sterile centrifuge tubes**Acceptable:** T-25 flasks of confluent cultured cells**Specimen Volume:** 20 mL**Specimen Stability Information:** Refrigerated (preferred)/Ambient**Specimen Type:** Chorionic villi**Container/Tube:****Preferred:** 15-mL tube containing 15 mL of transport media**Acceptable:** T-25 flasks of confluent cultured cells

Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Specimen Type: Blood spot
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: Perkin Elmer 226 (formerly Ahlstrom 226) filter paper, or Blood Spot Collection Card
Specimen Volume: 5 Blood spots

- Collection Instructions:**
1. An alternative blood collection option for a patient >1 year of age is a finger stick.
 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
 3. Do not expose specimen to heat or direct sunlight.
 4. Do not stack wet specimens.
 5. Keep specimen dry

Specimen Stability Information: Ambient (preferred)/Refrigerated

- Additional Information:**
1. For collection instructions, see [Blood Spot Collection Instructions](#) in Special Instructions.
 2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777) in Special Instructions.
 3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800) in Special Instructions.

- 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 in Special Instructions:
[-Informed Consent for Genetic Testing](#) (T576)
[-Informed Consent for Genetic Testing-Spanish](#) (T826)
 2. [Molecular Genetics: Congenital Inherited Diseases Patient Information](#) (T521) in Special Instructions.
 3. If not ordering electronically, complete, print, and send a [Neurology Specialty Testing Client Test Request](#) (T732) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
113452	Result Summary	Alphanumeric		50397-9
113453	Result	Alphanumeric		49857-6
113454	Interpretation	Alphanumeric		69047-9
113455	Additional Information	Alphanumeric		48767-8
113456	Specimen	Alphanumeric		31208-2
113457	Source	Alphanumeric		31208-2

113458	Released By	Alphanumeric		18771-6
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LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

- 81329
- 88235 (if appropriate)
- 88240 (if appropriate)
- 88233 (if appropriate)
- 88240 (if appropriate)
- 81265 (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
MATCC	Maternal Cell Contamination, B	1	81265	No	Yes
_STR1	Comp Analysis using STR (Bill only)	1	81265	No	No, (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)	1	81266	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
CULFB	52327	Result Summary	Alphanumeric		50397-9
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
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	53291	Released By	Alphanumeric		18771-6
MATCC	55150	Method	Alphanumeric		85069-3

Reference Values:

An interpretive report will be provided.