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**Reporting Title:** CYP21A2 Gene, Full Gene Analysis**Performing Location:** Rochester**Ordering Guidance:**

This test is a molecular analysis of the *CYP21A2* gene and does not include biochemical analysis of steroids. For biochemical analysis for congenital adrenal hyperplasia (CAH), which includes cortisol, androstenedione, and 17-hydroxyprogesterone, see CAH21 / Congenital Adrenal Hyperplasia (CAH) Profile for 21-Hydroxylase Deficiency, Serum.

**Additional Testing Requirements:**

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

**Shipping Instructions:**

Specimen preferred to arrive within 96 hours of collection.

**Necessary Information:**

[CYP21A2 Gene Testing for Congenital Adrenal Hyperplasia Patient Information](#) (T663) is strongly recommended, but not required, to be filled out and sent with the specimen. This information aids in providing a more thorough interpretation of test results. Ordering providers are strongly encouraged to complete the form and send it with the specimen.

**Specimen Requirements:**

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

**Submit only 1 of the following specimens:****Specimen Type:** Whole blood**Container/Tube:**

Preferred: Lavender top (EDTA)

**Specimen Volume:** 3 mL**Collection Instructions:**

1. Invert several times to mix blood.
2. Send specimen in original tube. **Do not** aliquot.

**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.**

**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.

**Specimen Type:** Chorionic villi  
**Container/Tube:** 15-mL tube containing 15 mL of transport media  
**Specimen Volume:** 20 mg  
**Specimen Stability Information:** Refrigerated

Additional Information:

1. 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.
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 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  
**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.

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. [CYP21A2 Gene Testing for Congenital Adrenal Hyperplasia Patient Information](#) (T663) is recommended.

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
37488	Result Summary	Alphanumeric		50397-9
37489	Result	Alphanumeric		82939-0
37490	Interpretation	Alphanumeric		69047-9

37491	Additional Information	Alphanumeric		48767-8
37492	Specimen	Alphanumeric		31208-2
37493	Source	Alphanumeric		31208-2
37494	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81405-CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (eg, steroid 21-hydroxylase isoform, congenital adrenal hyperplasia), full gene sequence

81402-CYP21A2 (cytochrome P450, family 21, subfamily A, polypeptide2) (eg, congenital adrenal hyperplasia, 21-hydroxylase deficiency), common variants (eg, IVS2-13G, P30L, I172N, exon 6 mutation cluster [I235N, V236E, M238K], V281L, L307FfsX6, Q318X, R356W, P453S, G110VfsX21, 30-kb deletion variant)

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

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (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)

81479 (if appropriate for government payers)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
MATCC	Maternal Cell Contamination, B	1	81265	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test	1	88235	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.