



# CYP21A2 Gene Testing for Congenital Adrenal Hyperplasia Patient Information



**Instructions:** Accurate interpretation and reporting of the genetic results is contingent upon the reason for testing, clinical information, ancestry, and family history. To help provide the best possible service, supply the information requested below and **send this paperwork with the specimen or return by fax to Mayo Clinic Laboratories, Attn: Molecular Genetics Lab Genetic Counselors at 507-284-1759. Phone: 800-533-1710 / International clients: +1-507-266-5700 or email MLIINT@mayo.edu**

### Patient Information (required)

Patient Name (Last, First Middle)		Birth Date (mm-dd-yyyy)
Sex Assigned at Birth <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Unknown <input type="checkbox"/> Choose not to disclose	Legal/Administrative Sex <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Nonbinary	

### Referring Provider Information

Requesting Provider Name (Last, First)	Phone	Fax*
Genetic Counselor Name (Last, First)	Phone	Fax*

\*Fax number given must be from a fax machine that complies with applicable HIPAA regulations.

For diagnostic testing or carrier screening on whole blood, order CYPZ / 21-Hydroxylase Gene CYP21A2, Full Gene Analysis, Varies.

- Note:**
- Due to the complexity of CYP21A2 testing, known variant testing is not available. If familial variants have been previously identified in this family, provide this information in the Family History section below and attach any available laboratory test reports from family members.
  - For prenatal specimens, order CYPZ / 21-Hydroxylase Gene CYP21A2, Full Gene Analysis, Varies.

### Reason for Testing

Diagnosis or Suspected Diagnosis (Indicate relevant information in the Clinical History section below.)

Prenatal (Indicate relevant information in the Clinical History section below.)

Carrier Screening:  Family history of the condition, specify: \_\_\_\_\_

Partner has a family history of the condition

Partner is a carrier of the condition

Partner is affected with the condition

Other reproductive risk assessment, specify: \_\_\_\_\_

### Pertinent Clinical and Laboratory History Check all that apply.

Suspected difference of sex development (such as clitoromegaly) detected on prenatal ultrasound

Positive newborn screen for CAH (Congenital Adrenal Hyperplasia)

Elevated 17-hydroxyprogesterone

Chromosome analysis performed, indicate patient's karyotype:  46, XX  46, XY  Other, specify: \_\_\_\_\_

History of salt-wasting

Precocious puberty or virilization, specify: \_\_\_\_\_

Other Information (eg, specific prenatal findings)

### Ancestry Ancestry may assist with interpretation of test results.

African/African American  East Asian  Latino/Latina  South Asian  Unknown

Ashkenazi Jewish  European  Middle Eastern  None of the above  Choose not to disclose

### Family History

Are other relatives known to be affected? <input type="checkbox"/> Yes <input type="checkbox"/> No	If "Yes," indicate their relationship to the patient.: _____
Are other relatives known to be a carrier? <input type="checkbox"/> Yes <input type="checkbox"/> No	If "Yes," indicate their relationship to the patient.: _____
Have other relatives had molecular genetic testing? <input type="checkbox"/> Yes <input type="checkbox"/> No	If "Yes," indicate familial variants and attach a copy of the family member's lab report.: _____
If the relative was tested at the Mayo Clinic, include the name of the family member: _____	

### New York State Patients: Informed Consent for Genetic Testing is required.

See Informed Consent for Genetic Testing (T576) or Informed Consent for Genetic Testing – Spanish (T826).