

CYP21A2 Gene Testing for Congenital Adrenal Hyperplasia Patient Information



**Instructions:** Accurate interpretation and reporting of the genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. To help provide the best possible service, supply the information requested below and **send a completed copy of this form with the specimen.** 

## Patient Information (required)

Patient Name (Last, First, Middle)	Birth Date (mm-dd-yyyy)	Sex
		🗆 Male 🛛 Female
Referring Provider Name (Last, First)	Phone	Fax*
Other Contact Name (Last, First)	Phone	Fax*
*Fax number given must be from a fax machine that complies with applicable HIPAA regulat		

For diagnostic testing or carrier screening on whole blood, order test code CYPZ / 21-Hydroxylase Gene (CYP21A2), Full Gene Analysis, Varies. **Note:** 

- Due to the complexity of *CYP21A2* testing, known mutation testing is not available. If familial mutations 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: \_
- $\hfill\square$  Partner has a family history of the condition
- $\hfill\square$  Partner is a carrier of the condition
- $\hfill\square$  Partner is affected with the condition

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

Ambiguous genitalia detected on prenatal ultrasound		
Positive newborn screen for CAH (Congenital Adrenal Hyperplasia)		
Elevated 17-0HP		
Chromosome analysis performed		
If yes, indicate patient's karyotype: 🗆 46, XX 🗆 46, XY 🔅 Other, specify:		
□ History of salt-wasting		
Female with pre- or post-natal virilization		
Male with childhood virilization		
Female with precocious puberty		
Other Information (eg, specific prenatal findings)		
Ethnic Background (Ethnic background may assist with interpretation of test results.)		
European/Caucasian; list countries of origin:		
🗆 African American 🗆 Hispanic 🗆 Asian 🗆 Middle Eastern 🗀 Other, specify:		

## **Family History**

Are other relatives known to be affected?	If yes, indicate their relationship to the patient.	
Are other relatives known to be a carrier?	If yes, indicate their relationship to the patient.	
🗆 Yes 🗆 No		
Have other relatives had molecular genetic testing?	If yes, indicate familial mutations and attach a copy of the family member's lab report.	
□ Yes □ No		
If the relative was tested at the Mayo Clinic, include the name of the family member:		