

The information below is required to perform genetic testing for congenital adrenal hyperplasia. Please complete this form and submit it with the test request form or electronic packing list.

CAH (21-Hydroxylase Deficiency) Common Mutations

Please complete the following:

Patient _____ Accession # _____ Date of birth _____

Date drawn _____ Ethnic background _____

Indication for test:

Positive newborn screen

Patient affected with:

Classic congenital adrenal hyperplasia (CAH)

Salt-wasting

Non-salt-wasting

Non-classic CAH

Ambiguous genitalia

Hirsutism

Polycystic ovarian syndrome

Other: _____

If affected, please provide biochemical test results, if performed:

Serum 17-OHP: _____

Plasma renin activity (PRA): _____

Stimulated ACTH: _____

Other (androstenedione, testosterone): _____

Patient has a positive family history of the following:

(Please detail affected individual's relationship to patient.)

Known familial mutation: _____

(Please indicate name of mutation(s); e.g., V281L)

Patient is having carrier screening because partner is a known carrier of CAH.