Quest Diagnostics

CAH (21-Hydroxylase Deficiency) Common Mutations

Please complete the following:

PATIENT: 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), ex: V281L)
- ❑ Patient is having carrier screening because partner is a known carrier of CAH

YOUR NAME AND PHONE NUMBER:_____

YOUR FAX NUMBER:_____

PLEASE FAX THIS FORM TO the Quest Genomics Testing Center at 610-271-4894. If you have questions, please call 866-436-3463 to speak with any Quest Diagnostics Genetic Counselor.