

T1 Key diagnostic steroids for different forms of classic CAH

| STEROID | HIGH IN | LOW IN |
|------------------------|--|---|
| Progesterone | 21OHD, 11OHD, 17OHD, PORD, 3 β HSD | LCAH |
| 17-Hydroxyprogesterone | 21OHD, 11OHD, PORD, 3 β HSD | LCAH, 17OHD |
| 11-Deoxycorticosterone | 11OHD, 17OHD, PORD | LCAH, 21OHD, 3 β HSD |
| Corticosterone | 17OHD, PORD | LCAH, 21OHD, 11OHD, 3 β HSD |
| 11-Deoxycortisol | 11OHD | LCAH, 21OHD, 17OHD, PORD, 3 β HSD |
| Dehydroepiandrosterone | 3 β HSD | LCAH, 17OHD, PORD, (21OHD, 11OHD) |
| Androstenedione | 21OHD, 11OHD, (3 β HSD) | LCAH, 17OHD, PORD |
| Testosterone | 21OHD, 11OHD, (3 β HSD) | LCAH, 17OHD, PORD |

21OHD, 21-hydroxylase deficiency; 11OHD, 11 β -hydroxylase deficiency; 17OHD, 17-hydroxylase deficiency; PORD, P450-oxidoreductase deficiency; 3 β HSD, 3 β -hydroxysteroid dehydrogenase type 2 deficiency; LCAH, lipoid congenital adrenal hyperplasia. Parentheses indicate variable or age- and sex-dependent variations

T2 Laboratory testing for diagnosis and management of classic 21OHD

| ANALYTE | PHYSIOLOGY | GOALS AND COMMENTS |
|------------------------------|--------------------------|---|
| Plasma renin | Volume status | Low to normal unless hypertensive |
| Sodium | Glucocorticoid | Normal |
| Potassium | Mineralocorticoid | Normal |
| Androstenedione | Adrenal dominant | Assess with testosterone |
| Testosterone | Total androgens | Normal; adrenal + gonadal |
| 17-Hydroxyprogesterone | HPA axis status | Moderately elevated; varies with dosing |
| Sex hormone-binding globulin | Androgen binding | Determines free fraction; estrogen raises |
| -FOR MEN | | |
| Luteinizing hormone | Gonadal axis | Normal; low if adrenal androgen excess |
| Follicle-stimulating hormone | Testis integrity | Normal; >25 IU/L poor fertility prognosis |
| Androstenedione/testosterone | Androgen sources | Ratio should be <0.5 |
| Semen analysis | Fertility | Normal |
| -FOR WOMEN | | |
| Progesterone | Adrenal vs corpus luteum | Normal in follicular phase for fertility |