Clinical Use

• Diagnosis and differential diagnosis of congenital adrenal hyperplasia (CAH)

Reference Range

Androstenedione
Cortisol
DHEA
Deoxycorticosterone
11-Deoxycortisol
17-OH pregnenolone
17-OH progesterone
Progesterone
Testosterone

*See individual assays for age-related reference ranges.

Interpretive Information

Results vary depending on the defect (21-OH, 11β-OH, 17α-OH, 17,20-lyase, 3β-HSD, StAR, or CYPOR deficiency). See Test Application and Interpretation, Disorders of Adrenal Function, Tables 6-8 for more information.

Clinical Background

Classic congenital adrenal hyperplasia (CAH) manifests in the neonatal period with impaired cortisol synthesis caused by genetic mutations that result in deficient activity of 1 of the adrenal biosynthetic enzymes. The most common form, accounting for 90% to 95% of CAH patients, is 21-hydroxylase (P-450 CYP21A2) deficiency; 11β-hydroxylase (P-450 CYP11B1) deficiency accounts for another 5% to 8%. 3β-Hydroxysteroid dehydrogenase (HSD3B2), aldosterone synthase (CYP11B2), steroid acute regulatory protein (StAR), and P-450 oxidoreductase (CYPOR) deficiencies constitute the remainder of cases in infancy. 17-hydroxylase deficiency usually presents at the time of puberty with hypertension and hypokalemia due to decreased 17α-hydroxylation of pregnenolone and progesterone in the adrenal and increased production of mineralocorticoids. Gonadal steroid production also is blocked, and LH and FSH levels are increased. Females have primary amenorrhea and absent sexual characteristics, and males present with complete pseudohermaphroditism (external female genitalia, no uterus, no fallopian tubes).

Method

• Extraction, radioimmunoassay (RIA)
• Extraction, chromatography, RIA
• Liquid chromatography, tandem mass spectrometry (LC/MS/MS)

Specimen Requirements

2.4 mL refrigerated serum
1.5 mL minimum

No additive red top tube
An early morning specimen is preferred.
Specify age and sex on test request form.