Clinical Use
• Diagnose aldosterone synthase deficiency

Reference Range
Androstenedione *
11-Deoxycortisol *
18-Hydroxycorticosterone *
17-Hydroxyprogesterone *
*See individual assays for age-related reference ranges.

Interpretive Information
18-OH corticosterone/aldosterone ratio* >40
*See Test Application and Interpretation, Disorders of Adrenal Function, Tables 6-8.

Clinical Background
Aldosterone synthase deficiency (CYP11B2, P450c11ase) is a rare form of CAH in which only aldosterone synthesis is affected. Two forms have been identified; type I is characterized by decreased, and type II by increased, 18-hydroxycorticosterone. Infants usually present in the neonatal period with failure to thrive, recurrent dehydration secondary to salt-wasting, decreased blood pressure, and acidosis.

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

Specimen Requirements
1.8 mL refrigerated serum
0.8 mL minimum
No additive red top tube
An early morning specimen is preferred.
Specify age and sex on test request form.