

Clinical Use

- Diagnose 3β-hydroxysteroid dehydrogenase deficiency
- Diagnose adrenal virilizing tumors

Related Assays


- 17-Hydroxypregnenolone
- DHEA
- DHEA-sulfate

Reference Range

	ng/dL	ng/dL
Men		10-200
Women		10-230
Children		
ACTH stimulation	Baseline	60 min
<1 y	10-140	49-360
1-5 y	10-48	34-135
6-12 y	15-45	39-105
Tanner II-III		
Males	10-45	58-110
Females	15-84	33-140
Tanner IV-V		
Males	11-50	37-150
Females	20-77	91-220

Pediatric data from *J Clin Endocrinol Metab.* 1992;75:1491-1496.

Interpretive Information

-  • Congenital adrenal hyperplasia
- Adrenal virilizing tumor
- Cushing's disease
- Normal children and adults after ACTH administration

Clinical Background

Normal steroid-producing cells of the adrenal glands and gonads synthesize various steroids from circulating cholesterol ester. This results in small amounts of precursors such as pregnenolone, a Δ⁵ C-21 steroid, leaking into the circulation. Most of the circulating pregnenolone is from the adrenal cortex, and levels are modestly increased after ACTH administration in normal children and adults.

This assay is useful in diagnosing 3β-hydroxysteroid dehydrogenase (3β-HSD) deficiency, an unusual form of congenital adrenal hyperplasia associated with blocked cortisol synthesis and increased levels of Δ⁵ steroids. After diagnosis of 3β-HSD deficiency, the test can evaluate glucocorticoid replacement therapy. The assay is also helpful in suggesting the presence of an adrenal virilizing tumor, since almost all of these tumors secrete large amounts of DHEA-sulfate and pregnenolone.

Pregnenolone levels are moderately increased in Cushing's disease due to ACTH-secreting pituitary adenoma or ectopic causes.

Method

- Extraction, chromatography, radioimmunoassay (RIA)
- Analytical sensitivity: 10 ng/dL

Specimen Requirements

4 mL refrigerated serum
 1.1 mL minimum
 No additive red top preferred
 SST red top acceptable