LabCorp offers a DNA test to identify deficiencies in the 21-hydroxylase gene (CYP21A2), the most common cause of congenital adrenal hyperplasia (CAH), a potentially lethal genetic disorder caused by a defect in adrenal steroid synthesis. The combination of low cortisol and aldosterone and excess androgen production can negatively affect childhood growth and development, with symptoms ranging from life-threatening salt wasting to virilization and precocious puberty. CAHDetx (500768) evaluates the CYP21A2 gene for mutations that account for approximately 90% to 95% of all CAH cases.

- CAHDetx detects the 12 most common small mutations and large gene deletions/conversions in CYP21A2
- Molecular genetic testing of CYP21A2 confirms biochemical findings and identifies carriers
- Utilizes highly-specific PCR and multiplex mini-sequencing technology
- Assay developed and validated based on published methods by in-house Ph.D. research and development team
- Assay performed at Endocrine Sciences, a member of LabCorp's Specialty Testing Group

Molecular testing for CYP21A2 gene mutations may be useful to:
- Rule out CAH in asymptomatic newborns with elevated 17-OHP screening results
- Confirm a genetic basis for 21-hydroxylase deficiency and/or CAH
- Predict the likely phenotype of a patient based on the mutations identified in the CYP21A2 gene

CYP21A2 gene organization (this is an approximate scale)

Nonclassic mutations include: P30L, V281L, P453S
Simple virilizing mutations include: I172N, In2G (A/C→G)
Salt wasting mutations include: In2G (A/C→G), Exon 6 cluster [I236N, V237E, M239K], R356W, G110Δ8nt, F306+1nt, Q318X, deletions, and large conversions
Note: 21-hydroxylase deficiency is an autosomal recessive disease and the phenotype of a patient with compound heterozygous mutations typically reflects the less severe mutation.
Scientific Expertise

- Industry-leading Endocrine Sciences laboratory with 40+ year history
- Extensive endocrine test menu including biochemical testing to detect CAH by HPLC/MS-MS methods, with complete normative data from premature infants to elderly adults
- Ph.D. and M.D. consultation available

Superior Service

- Endocrine hotline staffed by experienced client services representatives
- Comprehensive services for the endocrinology specialist
- Broad network of managed care health plans
- Flexible connectivity options for test ordering and result reporting
- Patient service centers accessible nationwide
- Courier and logistics services
- Local sales representation

Endocrine Sciences Direct Accounts

Please use the following code to order directly from the laboratory via Esoterix services: CAHDetx . . . 504006