LabCorp offers a DNA test to identify deficiencies in the 21-hydroxylase gene (CYP21A2), the most common cause of congenital adrenal hyperplasia (CAH)\(^1\), 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.\(^2\)

- CAHDetx detects the 12 most common small mutations\(^2\) 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\(^2,3\)
- Confirm a genetic basis for 21-hydroxylase deficiency and/or CAH\(^2\)
- Predict the likely phenotype of a patient based on the mutations identified in the CYP21A2 gene\(^1,3\)

**CYP21A2 gene organization**\(^3\) (this is an approximate scale)

1. P30L
2. G110\(\triangle8\)nt
3. I172N
4. I236N, V237E, M239K
5. V281L
6. F306+t
7. Q318X
8. R356W
9. P453S

- Nonclassic mutations include: P30L, V281L, P453S\(^2\)
- Simple virilizing mutations include: I172N, In2G (A/C\(\rightarrow\)G)\(^1\)
- Salt wasting mutations include: In2G (A/C\(\rightarrow\)G), Exon 6 cluster [I236N, V237E, M239K], R356W, G110\(\triangle8\)nt, F306+1nt, Q318X, deletions, and large conversions\(^1\)
- **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.\(^3\)
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**

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Test No | Test Name
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500768 | **Congenital Adrenal Hyperplasia (CAH) 21-Hydroxylase (CYP21) Mutation**

Visit the online Test Menu at www.LabCorp.com for full test information, including CPT codes and specimen collection requirements.

References