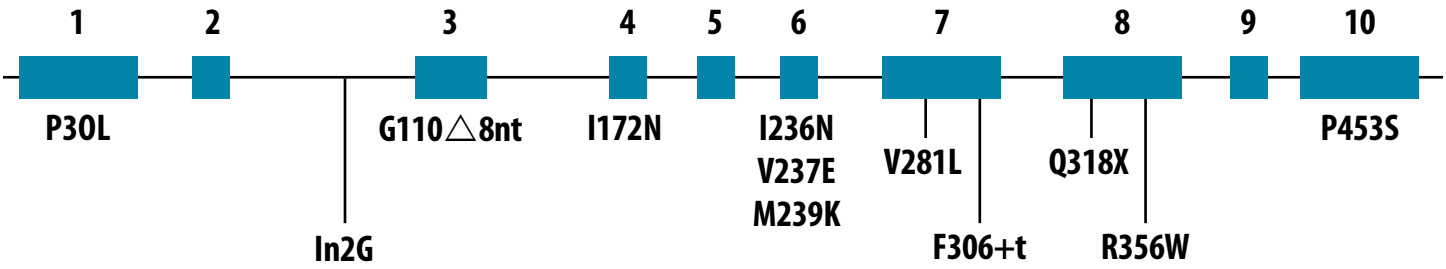
CAHDetx DNA TESTING
FOR 21-HYDROXYLASE DEFICIENCY

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)



- Nonclassic mutations include: P30L, V281L, P453S[2]
- Simple virilizing mutations include: I172N, In2G (A/C→G)[1]
- Salt wasting mutations include: In2G (A/C→G), Exon 6 cluster [I236N, V237E, M239K], R356W, G110Δ8nt, 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]

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Test No	Test Name
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.

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**

References

1. White PC and Speiser PW, 2000. Congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Endocr Rev* 21(3): 245-291. PubMed 10857554
2. Krone N, Braun A, Weinert S, et al., 2002. Multiplex minisequencing of the 21-hydroxylase gene as a rapid strategy to confirm congenital adrenal hyperplasia. *Clin Chem* 48(6): 818-825. PubMed 12028996
3. Speiser PW and White PC, 2003. Congenital adrenal hyperplasia. *N Engl J Med* 349: 776-788. PubMed 12930931

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