

DNA TESTING FOR 21-HYDROXYLASE DEFICIENCY

CAHDetx™

Esoterix introduces a new DNA test to identify deficiency in the 21-hydroxylase gene, the most common cause of congenital adrenal hyperplasia (CAH). CAHDetx evaluates the CYP21 gene, detecting mutations and gene deletion/conversions that account for approximately 90% to 95% of all CAH cases.

HIGH SCIENCE

- CAHDetx detects the 12 most common mutations and large gene deletion/conversions in CYP21 gene
- Molecular genetic testing of the CYP21 gene confirms the 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 Esoterix's specialized endocrinology molecular genetics laboratory

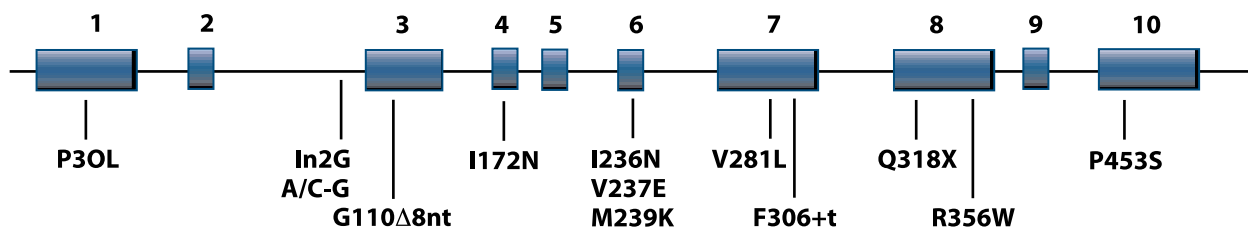
SUPERIOR CUSTOMER SERVICE

- Dedicated Ph.D. on staff to answer technical and clinical questions surrounding CAHDetx
- Dedicated courier and logistic services
- Local sales representation
- Client and third party billing options

INNOVATION

- Medtrax® – Internet ordering and reporting
- Entrix® – Direct interface connectivity with LIS
- Center for Innovation – Cutting edge research for the development of new assays

CYP21 Gene Organization (approximate scale only)



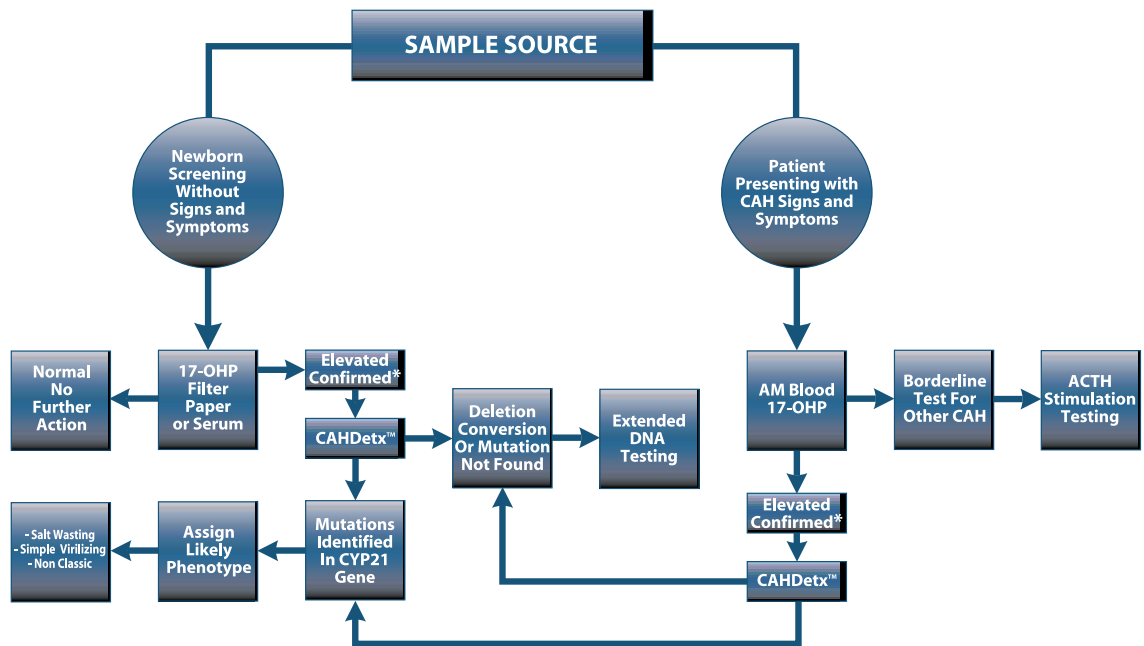
Nonclassic mutations: P30L, V281L.

Simple virilizing mutations: I172N, In2G (A/C-G).

Salt wasting mutations: In2G (A/C-G), 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 mutation typically reflects the less severe mutation.

CAHDetx™ Diagnostic Algorithm



*Monitor and treat patients clinically. If 17-OHP results are uncertain, repeat testing at Esoterix and do ACTH stimulation testing.

Test Code	504006
Specimen Requirement	3 mL whole blood [EDTA]. Ship at room temperature with cold pack preferred. Please include CAHDetx requisition if available.
Methodology	PCR, Multi-Plex Mini-Sequencing, fragment analysis
CPT Code	83891, 83898X3, 83894X6, 83892X2, 83901X3, 83912
Assay Time / Schedule	14 days Schedule As Needed

**For more information or to obtain a supply kit,
please call 800.444.9111 or visit www.esoterix.com.**

REFERENCES:

1. Speiser PW and White PC 2003. Congenital Adrenal Hyperplasia. *New England Journal of Medicine* 349:776-788.
2. Krone N, Braun A, Weinert S, Peter M, Roscher AA, Partsch C and Sippell WG 2002. Multiplex Minisequencing of the 21-Hydroxylase Gene as a Rapid Strategy to Confirm Congenital Adrenal Hyperplasia. *Clinical Chemistry* 48:818-825.
3. White PC and Speiser PW 2000. Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency. *Endocrine Reviews* 21:245-291.

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Laboratory Services
Anything But Routine™

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