Courtagen
DIAGNOSTICS LABORATORY

Courtagen's epiSEEK®

Comprehensive Sequence Analysis for Epilepsy and Seizure Disorders (327 genes)

Epilepsy is a common neurological disorder with a highly heterogeneous background and a strong genetic contribution.

The variety of non-specific and overlapping syndromic and nonsyndromic phenotypes can often hamper a clear clinical diagnosis, and prevents targeted genetic testing for selected mutations. In many cases, the precise genetic diagnosis can be important in therapy selection, particularly when there are know contraindications, such as with the prescription drug valproic acid.

Knowing the genetic basis of a patient's epilepsy is valuable for obtaining a definitive diagnosis, estimating prognosis, guiding treatment choices, and determining recurrence risks.

Courtagen Diagnostics Laboratory's epiSEEK® test provides the complete sequence of 327 genes that have been reported in association with epileptic and seizure disorder phenotypes. The gene categories are listed below.

Gene Categories

- Syndromic Disorders (71 genes)
- Brain or Nervous System Malformations (50 genes)
- Inherited Metabolic Diseases (41 genes)
- Neurodegeneration (32 genes)
- Idiopathic Generalized Epilepsy (28 genes)
- Glycosylation Disorders (23 genes)
- Mitochondrial Dysfunction (27 genes)
- Epilepsy in X-linked Intellectual Disability (18 genes)
- Early Infantile Encephalopathy (16 genes)
- Joubert Syndrome (9 genes)
- Other (12 genes)

Courtagen provides saliva DNA specimen collection kits at no charge.
Questions? Contact our Patient Advocates.

P 877.395.7608 | F 617.892.7192 | email genomics@courtagen.com. | www.courtagen.com

Performance

This test was designed to sequence the exons and canonical splice sites (+/-1,2) of a panel of genes associated with epilepsy and seizure disorders. Genomic DNA is extracted from the sample (typically saliva), submitted, and captured with an inversion probe method for the genes specific to this panel. The captured targets are sequenced on the Illumina MiSeq sequencing system with 250bp paired-end reads. Panel specific sequencing coverage, specificity, and sensitivity are listed below:

Mean depth of coverage:	500
Specificity:	99.99%
Sensitivity:	98.59%

Courtagen's Unmatched Customer Support

Turn Around Time: 4-6 weeks. Results are delivered in weeks, not months.

Saliva Sample: DNA for sequencing is reliably extracted from a single saliva sample. No blood draw or muscle biopsy required. (Blood and tissue are accepted, as requested.)

Insurance Assistance: Courtagen works with patients, physicians, and insurance carriers to pre-approve each test. Courtagen will bill the insurance company and is willing to handle an appeal process as needed.

Courtagen Care Financial Program: For qualified patients, the Courtagen Care Financial Program can help limit out–of–pocket expenses to $0, $50, or $100 per test.

Online Portal: A secure physician online portal is available for ordering genetic tests and accessing patient reports when completed.

Genetic Counselors: Available to address your questions regarding Courtagen test results.

Clinical Experience: Courtagen's Medical Director, Laboratory Director, and variant science team have over 25 years of experience in the treatment and genetic interpretation of neurological and metabolic disorders.

Reports: Utilizing Courtagen's customized Zypher® informatics pipeline and thorough clinical evaluation, each report is provided in a concise format with interpretation and recommendations for consideration.

Required forms

Courtagen Test Requisition (completed and signed by ordering clinician)

Signed Patient Consent (located on the Courtagen Test Requisition or Consent Form provided in the saliva kit)

Photocopy front & back of all insurance cards, including subscriber date of birth