Courtagen
DIAGNOSTICS LABORATORY

Courtagen's epiSEEK®

Sequence Analysis for Epilepsy and Seizure Disorders

The most complete tests available for seizure disorders

Knowing the genetic basis of a patient's epilepsy is valuable for obtaining a definitive diagnosis, estimating prognosis, determining recurrence risks, and guiding treatment choices. In many cases, the precise genetic diagnosis can be important in therapy selection, particularly when there are known contraindications or recommended treatment options based on genetic results.

epiSEEK® Triome™ (1,089 genes): Designed to provide the most complete picture, the epiSEEK Triome panel uses powerful trio analysis, by fully sequencing the parent's samples along with the patient, to enable extensive genetic analysis and clinical interpretation of an expanded selection of genes associated with neurological function, and epileptic and seizure disorder phenotypes. Unique features include:

- Drug metabolism guidance for 24 classes of anti-epileptic drugs and associated interactions.
- Includes genes from epiSEEK Comprehensive

epiSEEK® Comprehensive (471 genes): Designed to provide a thorough view, the epiSEEK Comprehensive panel enables genetic analysis and clinical interpretation of a wide range of genes associated with neurological function, and epileptic and seizure disorder phenotypes. Unique features include:

- Drug metabolism guidance for 24 classes of anti-epileptic drugs and associated interactions.
- Genes for other disorders and pathways are included to aid in differential diagnosis, such as inborn errors of metabolism, congenital disorders of glycosylation, peroxisomal biogenesis disorders, seizures related to intellectual disability, and cannabinoid pathway genes.

epiSEEK® Focus (76 genes): Designed to target the most common disease associated genes related to epileptic and seizure disorders – does not include drug metabolism.

Del/Dup Additional Analysis (76 genes): Additional analysis of exon-sized (and larger) deletion and duplication variants in 76 genes is available as an add-on for the epiSEEK Focus and epiSEEK Comprehensive panels only.

Indications for Testing

Primary Indications
Seizures
Infantile spasms
EEG abnormality
Brain malformations
Epileptic Encephalopathy

Secondary Indications (examples)
Autism spectrum disorder
Developmental delay
MRI abnormalities
Neuroregression
Cognitive impairment
Visual impairment
Movement disorders
Ataxia

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 of the genes associated with epilepsy and seizure disorders. Genomic DNA is extracted from the submitted sample (typically saliva), 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:

	epiSEEK Triome	**epiSEEK Comprehensive**	**epiSEEK Focus**
Mean depth of coverage:	>500	>500	>500
Target region covered:	99.5%	98.9%	98.2%
Quality threshold:	98.3%	96.8%	96.0%

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; however blood and muscle tissue are accepted. No charge saliva kits are provided, no charge phlebotomy services are offered.

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 Assistance Program: Provides financial assistance to patients based on demonstrated financial need. A qualified applicant may be asked to contribute between $0 and a maximum cost sharing of $200 depending on determination of financial need.

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.
Contact us at clinical@courtagen.com.

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 ZiPhyr® informatics pipeline and thorough clinical evaluation, each report is provided in a concise format with interpretation and recommendations for consideration.

Data and DNA Security: Advanced IT solutions safeguard patient records and financial information. In the laboratory, Courtagen de-identifies patient and test records and uses the proprietary method of DREAM PCR to provide exceptional lab decontamination.

Required forms

Courtagen Test Requisition (completed and signed by ordering clinician)

Signed Patient Consent (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)