Courtagen
DIAGNOSTICS LABORATORY

Courtagen's nucSEEK®

Mitochondrial Disease

DNA Sequence Analysis of Nuclear Mitochondrial Genes

Genes in the nuclear mitochondrial exome continue to surface as significant factors in disease

Mitochondrial disorders are very diverse conditions that can affect multiple systems of the body. Mitochondrial dysfunction depletes cells of energy causing cell damage and even cell death. Due to the high energy requirements of brain and muscle, mitochondrial disease may present more prominently in these areas. Other organs frequently affected include eye, ear, heart, liver, kidney, gastrointestinal tract, nervous system, and endocrine organs. The analysis and interpretation of a patient's nuclear mitochondrial genes using Next Generation Sequencing is valuable for obtaining a definitive diagnosis, estimating prognosis, guiding treatment choices, and determining recurrence risks.

nucSEEK® Comprehensive (1,189 genes): Over 1,000 proteins encoded in the nuclear genome play a role in mitochondrial processes. Courtagen's nucSEEK Comprehensive test detects variants in the full range of functionally associated genes to provide the complete picture of the nuclear mitochondrial exome.

nucSEEK® Focus (181 genes): Designed to target the documented disease associated nuclear genes related to known mitochondrial disorders.

Del/Dup Additional Analysis (181 genes): Additional analysis of exon-sized (and larger) deletion and duplication variants in 181 genes is available as an add-on for both the nucSEEK Focus and nucSEEK Comprehensive panels.

Indications for Testing

- Seizures
- Ataxia
- Early-onset stroke
- Retinopathy
- Sensorineural hearing loss
- Ophthalmoplegia
- Ptosis
- Migraine
- Chronic pain syndromes
- Fatigue
- Gastrointestinal dysmotility
- Dizziness/POTS, arrhythmia
- Depression
- Anxiety
- Cognitive
- Intellectual disability
- Autistic spectrum disorders
- ADHD
- Muscular weakness
- Cardiomyopathy
- Endocrinopathies
- Pancytopenias
- Exocrine pancreatitis
- Liver failure
- Renal tuberopathy

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

These tests are designed to sequence the exons and canonical splice sites of the nuclear encoded mitochondrial genes as listed by MitoCarta at the time of development. Genes were included in the panel if they represented a phenotypically similar disease (e.g. *MECP2* for Rett Syndrome). 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:

	nucSEEK Comprehensive	nucSEEK Focus
Mean depth of coverage:	>400	>400
Target region covered:	99.7%	99.9%
Quality threshold:	98.6%	98.7%

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. Immediate results can also be faxed.

Genetic Counselors: Available to address physician's 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)