Courtagen
DIAGNOSTICS LABORATORY

Courtagen's mtSEEK®

Mitochondrial Disease

Mitochondrial Genome Sequencing with Heteroplasmy Analysis

Courtagen's mtSEEK® test provides results from saliva and eliminates the need for a costly, invasive, and risky muscle biopsy.

Mitochondrial disorders are very diverse conditions and can affect one or multiple systems of the body. Mitochondrial dysfunction depletes cells of energy, causing cell damage and even cell death. Diseases of the mitochondria appear to cause the most damage to cells of the brain, heart, liver, skeletal muscles, kidney and the endocrine and respiratory systems.

Courtagen's mtSEEK® test uses Next Generation Sequencing to sequence the 16,569 bases that make up the 37 genes of the mitochondrial genome. The proprietary assay design provides contamination control, highly accurate mapping and sensitive detection of mtDNA heteroplasmy levels as low as 5%.

Common Symptoms

Brain

Developmental delays, dementia, migraines, autistic features, intellectual disability, seizures

Nervous System

Weakness, fainting, dizziness, neuropathic pain, heat or cold intolerance, pallor

Muscles

Weakness, fatigue, pain, cramping, exercise intolerance

Liver and Kidneys

Liver failure, hypoglycemia (low blood sugar), renal tubular acidosis or wasting

Gastrointestinal

Dysmotility, diarrhea or constipation, cyclic vomiting, failure to thrive, gastroesophogeal reflux

Heart

Cardiomyopathy, cardiac conduction defects

Ears and Eyes

Visual loss, ptosis (droopy eyelids), opthalmoplegia, optic atrophy, hearing loss

Endocrine

Diabetes, exocrine pancreatic failure, parathyroid failure (low calcium)

Mitochondrial disorders which are caused by mutations in the nuclear mitochondrial DNA are not assayed by this test. The nuclear mitochondrial exome (approx.1,200 genes) is available with the Courtagen nucSEEK® Comprehensive panel.

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

Accurate variant detection. mtDNA is amplified using long range PCR, and Next Generation Sequencing is performed with 250bp paired-end reads – providing highly accurate mapping and assembly, and avoiding nuclear mtDNA (numt) mismapping.

Sensitive heteroplasmy detection. The use of deep sequencing coverage delivers maximum sensitivity for heteroplasmy detection as low as 5%.

Contamination control. Courtagen's proprietary D.R.E.A.M PCR technique results in exceptional laboratory decontamination, critical for sensitive heteroplasmy detection.

Mean depth of coverage:	> 2,000
Specificity:	>99%
Sensitivity:	>99%

Courtagen's Unmatched Customer Support

Turn Around Time: 3-5 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, and 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 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.

Required forms

Courtagen Test Requisition (completed and signed by ordering clinician)

Signed Patient Consent (located on Consent Form in the saliva kit)

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