Courtagen
DIAGNOSTICS LABORATORY

Courtagen's nucSEEK®

Mitochondrial Disease

Comprehensive Sequence Analysis of the Nuclear Mitochondrial Exome (1,100+ genes)

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

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. Due to the high energy requirement of brain and muscle, mitochondrial disease typically affects these parts of the body. Other organs frequently affected include eye, ear, heart, liver, kidney, gastrointestinal tract, nervous system, and endocrine organs.

There are over 1,000 proteins encoded in the nuclear genome that are imported into the mitochondria. Courtagen's nucSEEK® test uses Next Generation Sequencing to detect variants in these nuclear genes. The analysis and interpretation of a patient's nuclear mitochondrial exome is valuable for obtaining a definitive diagnosis, estimating prognosis, guiding treatment choices, and determining recurrence risks.

Gene Categories

- ABC Transporters (14 genes)
- Amino Acid Metabolism (48 genes)
- Antioxidant Pathways (22 genes)
- Carbohydrate Metabolism (36 genes)
- Cell Function (118 genes)
- Complex I (52 genes)
- Complex II (6 genes)
- Complex III (10 genes)
- Complex IV (32 genes)
- Complex V (23 genes)
- Cofactors (25 genes)
- Dynamics (9 genes)
- DNA/RNA (32 genes)
- GTPase (20 genes)
- Ion Channel (31 genes)
- Iron Metabolism (105 genes)
- Membrane Protein Transport (20 genes)
- Neurotransmitter (17 genes)
- Nucleotide Metabolism (17 genes)
- Other (180 genes)
- Other Enzyme (91 genes)
- Peroxisome (18 genes)
- Protein Metabolism (32 genes)
- Ribosome Protein (84 genes)
- Solute Carrier (56 genes)
- TCA Cycle (35 genes)
- tRNA (39 genes)

Performance

This test was designed to sequence the exons and splice sites (+/-10) 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:

Mean depth of coverage:	500
Specificity:	99.99%
Sensitivity:	99%

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

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