

## Courtagen's lysoSEEK™



# Lysosomal Storage Disorder Panel (94 genes)

**Next Generation Sequencing test focused on early identification of genes associated with potentially treatable storage disorders**

Lysosomal storage disorders are part of the differential diagnosis of several, diverse perinatal and childhood phenotypes, which are extremely variable in both onset and clinical severity. Prompt diagnosis (within the first year), including genetic testing, may enable early treatment (e.g. enzyme replacement therapy), possibly preventing irreversible clinical consequences.

Courtagen's lysoSEEK panel provides extensive genetic analysis and clinical interpretation of data generated by the complete sequencing of 94 genes associated with over 60 metabolic disorders (e.g. Tay-Sachs, Neimann-Pick, Walker-Warburg, Fabry, and others). The test seeks out enzymatic deficiencies in the lysosomes and peroxisomes, and looks at additional genes useful for differential diagnoses.

### Indications for Testing

- Failure to thrive
- Developmental delay
- Developmental regression
- Seizures
- Recurrent respiratory infections
- Endocrine Dysfunction
- Cardiovascular abnormalities
- Facial dysmorphism
- Hepatosplenomegaly
- Corneal clouding
- Oral findings (e.g. macroglossia, molar hypoplasia, hypertrophic gums)
- Stiff joints
- Dysostosis multiplex
- Exaggerated startle
- *Other Indications*

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](mailto:genomics@courtagen.com) | [www.courtagen.com](http://www.courtagen.com)

## Performance

This test was designed to sequence the exons and canonical splice sites (+/-1,2) of a panel of 94 genes associated with lysosomal storage and peroxisomal biogenesis 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:

Mean depth of coverage:	300
Specificity:	99.99%
Sensitivity:	98%

## 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 also 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 physician's questions regarding Courtagen test results. Contact us at [clinical@courtagen.com](mailto: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)