Courtagen
DIAGNOSTICS LABORATORY

Courtagen's devACT®

Neuro-development

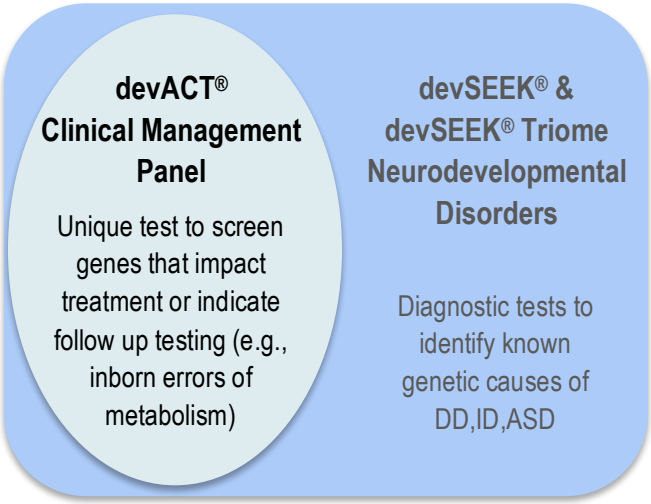
Focus on Actionable Results

Unique Next Generation Sequencing test that informs *actionable clinical management* of patients presenting with Developmental Delay, Intellectual Disability, and/or Autism Spectrum Disorders

Courtagen's devACT® Clinical Management Panel provides an efficient screen of over 250 genes that can inform **meaningful treatment options** while potentially avoiding both costly and invasive alternative diagnostic procedures.

Many patients presenting with developmental delay (DD), intellectual disability (ID), and/or autism spectrum disorders (ASD), experience a diagnostic work-up that consists of many tests over an extended time period. Results from Courtagen's devACT Clinical Management Panel can be obtained from a single saliva sample and enables an efficient sequencing screen for **treatable genetic conditions** in patients presenting with clinical features of DD, ID, and/or ASD. This panel promotes treatability in the work up of patients with the potential to improve health outcomes.

The devACT panel can be combined with the devSEEK or devSEEK Triome panel for additional analysis of syndromic and non-syndromic causes of DD, ID, and ASD.



Effectively screen for many conditions

devACT is a sequencing panel that includes treatable causes of ID/DD/ASD, as well as tests for disorders that are currently queried for by extensive biochemical testing, such as Congenital Disorders of Glycosylation (CDG), Very Long Chain Fatty Acids (VLCFA), oligosaccharides/mucopolysaccharidoses (MPS), Urine Organic Acids (UOA), Plasma Amino Acids (PAA), neurotransmitters, pterins, folate metabolites, cerebrospinal fluid (CSF) glucose, magnetic resonance spectroscopy (MRS) for creatine, purines and pyrimidines, etc. The devACT panel is designed to be indispensable as it screens for many treatable conditions far more effectively than lengthy diagnostic work ups that are substantially less complete. Additional analysis of exon-sized (and larger) deletion and duplication variants in 20 genes is available as an add-on.

Potential Procedures to Avoid

Anesthesia, Biopsy, Lumbar Puncture, MRI, Multi-test work up over extended period

Potential Treatments

The goal of this genetic test to is to identify and prioritize treatability. Therapeutic modalities may include: sick-day management, diet, co-factor/vitamin supplements, substrate inhibition, stem cell transplant, gene therapy [1]. Examples include: Biotin Therapy, Creatine Therapy, Folinic Acid, CoQ10, L-carnitine, Riboflavin, Vitamin B_6, Vitamin B_{12}, Vitamin K, Ketogenic diet

[1] C.D.M. van Karnebeek, S. Stockler, Treatable inborn errors of metabolism causing intellectual disability, Mol. Genet. Metab. (2012)

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

Test Overview

This test was designed to sequence the exons and canonical splice sites (+/-1,2) of a panel of 255 genes that can inform meaningful treatment options while potentially avoiding costly and invasive diagnostic procedures associated with developmental delay, intellectual disability and autism spectrum 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:	>400
Specificity:	99%
Sensitivity:	98.3%

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 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)