Welcome to the new era of genomics-based healthcare.

Introducing TruGenome Clinical Sequencing Services from Illumina Clinical Services Laboratory
Genomics-based healthcare is available now for you and your patients.

We are in the beginning of a new era of using genomic information to make critical healthcare decisions. To offer you and your patients all the advantages of genetic analysis technologies, Illumina Clinical Services Laboratory provides the most advanced solution in genomics-based healthcare: TruGenome Sequencing.

TruGenome Clinical Sequencing Services helps you both diagnose and assess your patients’ risk for certain genetic diseases.

- With whole-genome sequencing, we can deliver comprehensive answers—accurately and quickly—to your most challenging questions about genetic aberrations and rare diseases.
- You can make genetically informed decisions personalized to each patient.
- Ultimately, genomics-based healthcare may enable us to manage health through wellness, not illness.

Whole-genome sequencing is proving to be an invaluable tool in the identification of rare and undiagnosed disease, and as we learn more about the human genome and its impact on human health, I expect sequencing to become a regular component of healthcare. Medical College of Wisconsin’s partnership with Illumina for clinical testing services has greatly advanced our capacity in this area.

- Howard Jacob, Director of the Human and Molecular Genetics Center and Warren P. Knowles Chair of Genetics at the Medical College of Wisconsin.
The first CLIA-certified, CAP-accredited laboratory protocol

Since 2001, Illumina has been the research community’s partner of choice for innovations in DNA analysis technology.

In 2009, Illumina Clinical Services Laboratory became the first CLIA-certified, CAP-accredited lab to offer easy access to human whole-genome sequencing services. By providing the technology and interpretation expertise and tools to perform genomics-based testing, Illumina is paving the way for more clinical laboratories to offer these services worldwide.

A single test for comprehensive evaluation, today and tomorrow

Innovative Illumina technology enables genome-wide evaluation with just one test.

- We use the industry-leading, most widely published, next-generation sequencing technology to ensure results you can trust.
- With the most extensive experience interpreting whole-genome data, we provide comprehensive variant classifications based on known clinical relevance.
- Sequence today, plan for tomorrow: Our service ensures the capability for reanalysis in the future to keep pace with new discoveries.

Unsurpassed power to capture true biology

Offerings from the Illumina Clinical Services Laboratory—tailored to each patient’s condition and needs—focus on three major areas:

- **TruGenome Undiagnosed Disease Test**  
  For your patients with a suspected genetic disease who have gone through traditional genetic testing without a definitive diagnosis, we test for inherited diseases of single-gene etiology, such as mitochondrial, inherited cardiomyopathies, as well as multiple-symptom conditions, including seizures and degenerative neurogenetic conditions.

- **TruGenome Predisposition Screen**  
  For your healthy adult patients interested in learning about their carrier status or risk for adult-onset conditions, we perform sequencing and interpretation to analyze potential future risk for Mendelian (single-gene) conditions.

- **TruGenome Technical Sequence Data**  
  For labs and physicians performing their own clinical interpretations, we do all the sequencing using our CLIA-certified protocols.
See how simple it is to use TruGenome Clinical Sequencing Services
Physician–Patient Discussion
You discuss whole-genome sequencing with your patient and obtain formal consent. (Genetic counseling is also recommended.)

Physician–Illumina Clinical Geneticist Consultation
You place the order, request a sample kit, and consult with an Illumina Clinical Geneticist, depending upon the service selected.

Sample Collection
In your office or a reference lab, the patient’s blood sample is collected and then shipped to us.

Whole-genome sequencing at Illumina Clinical Services Laboratory
At our CLIA-certified, CAP-accredited lab, the DNA is sequenced, and variants are identified and classified. Our bioinformaticians, medical geneticists, and genetic counselors analyze and interpret the relevant gene variants.

Report Sent to Physician
We send you a report containing the clinical classifications of the variants found. (A clinical report is not provided with the Technical service.)

Physician–Patient Discussion
You use this report to discuss the results and next steps with your patient.
Accurate interpretation and clear reporting

The TruGenome clinical report includes:

- Summary
- Tables highlighting the number and category of variants clinically significant for the patient or for carrier status
- Brief description of the aberration or disease associated with the variant
- Explanation of the basis for our interpretation of the variant
- Information regarding relevant papers on the variant and allele frequency
- Information regarding the sequencing test itself
- Recommendations for follow up
- Appendix of all variants deemed to be benign, likely benign, or of uncertain significance

Six standards for clinical classification of variants

Pathogenic:
Segregates with disease in > 3 unrelated cases with control data. Located in a highly conserved region. Functional studies or other evidence suggest deleterious effect on gene expression.

Likely Pathogenic:
Reported in a few case studies with or without control data. Functional studies or other evidence suggest deleterious effect on gene expression.

Unknown Significance—Suspicious*:
Reported in a single or very limited number of cases, weakly suggestive of pathogenicity.

Unknown Significance:
Nothing has been reported regarding this variant, or reported information is incomplete and/or contradictory.

Likely Benign:
Reported in few cases, with little or no control data. May be non-conserved and/or predicted to be well tolerated. Frequency is higher than expected in the general population based on disease prevalence and penetrance. Functionally normal.

Benign:
Reported at high frequency in control data/does not segregate with disease. Variant may be non-conserved and/or predicted to be well tolerated. Functionally normal.

* Unique to Illumina.
TruGenome Clinical Sequencing Services

From Illumina Clinical Services Laboratory

www.illumina.com/clinicallab
You can start using TruGenome Clinical Sequencing Services today.

Welcome to the new era of genomics-based healthcare. With TruGenome Clinical Sequencing Services. Only from the Illumina Clinical Services Laboratory.

Learn more at www.Illumina.com/clinicallab