



Variantyx Lab Services

Enabling commercial and hospital labs to rapidly launch NGS tests





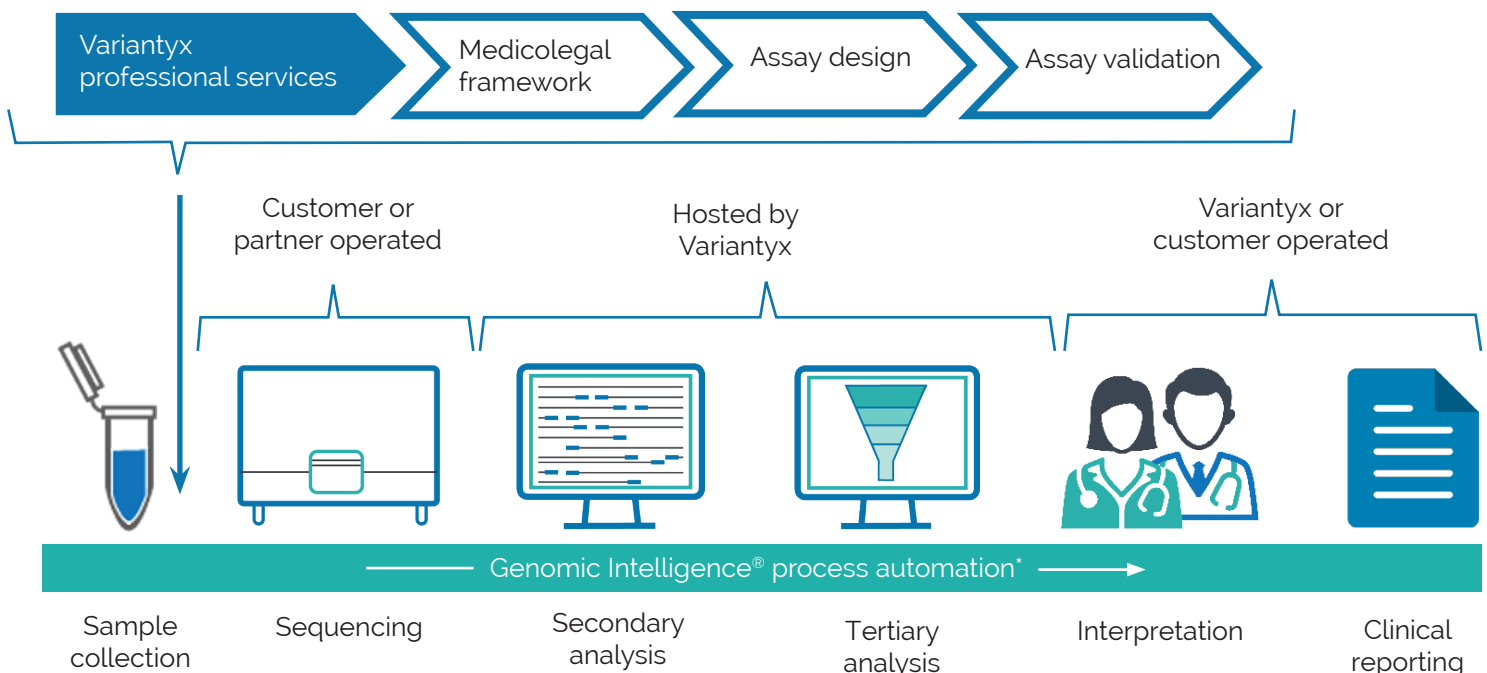
Variantyx is the leading provider of white-label genomic diagnostic solutions. We enable commercial and hospital labs to create profitable genetic tests, provided under their own brand, with minimal R&D investment. We specialize in four key application areas: rare inherited disease, cancer predisposition, prenatal and carrier screening.

Why Variantyx?

Whether you're already running NGS tests in your lab or are looking to upgrade Sanger sequencing tests to new NGS methods, you're probably familiar with the challenges of launching a new test. You need to design, develop, and validate your assay. You need to have an appropriate regulatory framework in place. In addition to sequencing, you need an integrated and automated data analysis pipeline, the ability to interpret variants, as well as expertise and authority to sign off on clinical reports.

As a clinically accredited diagnostic lab, we know these challenges first hand. Our clinically validated Genomic Intelligence® platform is used daily to process whole genome sequence (WGS) data for diagnosis of rare inherited disorders by our board-certified clinical and molecular geneticists. The platform simultaneously processes data from genomic tests of all sizes, from amplicon-based single gene assays to whole exome sequencing (WES) and WGS assays. With Variantyx Lab Services, we help labs like yours benefit from the tools and processes we have built and validated, as well as from our expertise in clinical interpretation and collaborative diagnosis.

The Genomic Intelligence® platform



*Purchase or license only the required components.

Powerful data QC, analysis and annotation tools

The Genomic Intelligence® platform is built to handle complex WGS data, automating the secondary and tertiary analysis process for data output of all sizes. The algorithms developed for our bioinformatic pipeline identify four different variant types:



Small sequence changes	Complete coverage of SNVs and small indels within coding and non-coding regions
Structural variants	Identification and annotation of large structural variants, including CNVs
Short tandem repeats*	Detection of short tandem repeats in known disease causing genes
Mitochondrial variants*	Detection of mitochondrial variants, including mitochondrial heteroplasmy down to 2%

* Requires WGS data

Relevant components are deployed to deliver amplicon-based tests, multigene panel assays, exome, and genome assays.

Genomic Intelligence® secondary and tertiary analysis features include:

- ✓ Extensive data QC including determination of mapped read percentage, coverage statistics, and pedigree concordance
- ✓ Best-in-class public and commercial variant and gene annotation databases
- ✓ *In silico* panels
- ✓ Population allele frequency databases
- ✓ Variant severity and conservation prediction tools
- ✓ Proprietary mode of inheritance data and phenotype-disease-genotype mapping algorithm



Interpretation and clinical reporting

Variantix clinicians use the cloud-based Genomic Intelligence® diagnostic console to review and interpret variants as well as generate and sign off on clinical reports.



An experienced partner in professional services

As a CLIA-certified diagnostic lab, we've clinically validated all components of the NGS testing process. We use our repository of true positive and true negative samples collected through the experience of launching our own high complexity testing to verify that your assay performs as expected. We will work with your team as needed to bring your assay to the required sensitivity, specificity, and positive predictive value (PPV) thresholds, benchmarking your assay's performance against our internal database of 5,000+ processed samples.

Unsure of the best assay design to meet your goals? Our expert R&D scientists can work with you to determine which type of assay to perform (panel, exome, etc), which genes to include, which primers or pull-down kit to use and which mutation types (SNVs, deletions/duplications, etc) to support. Our library of medicolegal templates ranging from test requisition forms (TRFs) and consent forms to clinical reports enables quick and easy customization.

Contact us today to start the conversation:
617-209-2090
[variantix.com/company/contact-us](https://www.variantix.com/company/contact-us)