Genomic Intelligence®

Providing higher diagnostic yield with fast turnaround time at a lower overall cost

Variantyx combines advanced technology with the framework of a clinical diagnostic lab to provide clinically validated solutions that enable labs to easily expand their test menu.

Genomic Intelligence® is an automated end-to-end platform that simplifies NGS data analysis, interpretation and clinical reporting. All aspects of the NGS testing process are seamlessly integrated to deliver the highest level of operational efficiency coupled with superior diagnostic results.

Components include:

- Sample collection and test selection
- DNA extraction, QC and sequencing
- Data QC, analysis and annotation
- Interpretation and clinical reporting

Genomic Intelligence® has been tested and validated as a clinical grade platform by our in-house board-certified medical geneticists as well as third parties who have used it to evaluate more than 4,000 samples to date.

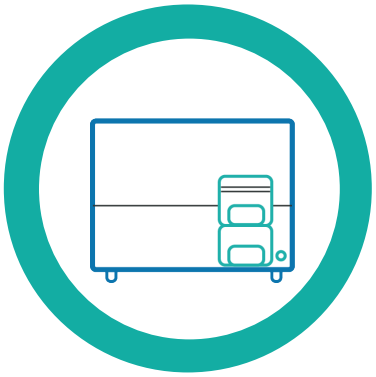
Genomic Intelligence® is licensed as a complete end-to-end platform. The analysis and reporting components can be licensed separately if desired.

Sample collection and test selection

Prepackaged sample collection kits with unique barcodes make it easy to initiate the testing process while maintaining:

- Patient privacy
- Full regulatory compliance
- Custody chain audit trail
- Tracking of sample progress

Ordering physicians have full flexibility to select from panel, whole exome or whole genome testing options for the patient plus up to six relatives. And to provide input on the patient's phenotype to guide the analysis.



DNA extraction, QC and sequencing

Integration with world-leading CLIA-certified and CAP-accredited sequencing partners ensures the highest standards of DNA extraction and sequencing. As well as an unbroken audit trail and ongoing oversight of the sample's progress at all times. Pre-negotiated discounts guarantee competitive per-sample pricing with no minimum volume required. Custom LIMS integration is available as an alternative.

Data QC, analysis and annotation

FASTQ data is securely transferred to Variantyx's cloud-based, HIPAA-compliant platform. The bioinformatic pipeline provides rapid, accurate analysis and annotation employing:

- Extensive data QC including determination of mapped read percentage, coverage statistics, gender and pedigree concordance
- Gold-standard algorithms and best practices for alignment and variant calling
- Proprietary methods for calling structural variants, trinucleotide repeat expansions and mitochondrial variants
- Best-in-class public and commercial variant and gene annotation databases
- Multiple population allele frequency databases
- Proprietary mode of inheritance data and phenotype-disease-genotype mapping algorithm
- Widely accepted variant severity and conservation prediction tools
- In silico panels

Variantyx's multi-site, fully redundant data centers guarantee the highest levels of data retention, security and compliance.

Interpretation and clinical reporting

Analyzed and annotated data are automatically transferred to your clinical team for expert review. The Genomic Intelligence® diagnostic console enables:

- Evaluation of variants against known patient phenotypes
- Links to supporting literature for annotated variants
- Storage of your lab's variant findings and comments
- One click generation of a white labeled clinical report
- Optional automated reanalysis of cases

Our board-certified medical geneticists are available to help make a collaborative diagnosis or to provide a second opinion.

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