Human Genomics

SURVEY OF CLINICALLY RELEVANT VARIANTS

IGATech™

IGATech
SERVING HUNGRY SCIENTISTS

IGATech provides full support on study design to ensure correct sequencing and bioinformatics strategies are used to meet your project goals. Our experts will consult with you about your specific requirements being your reference contact for the length of your project.

Tackle complex biological questions and improve diagnostics and routine clinical care.

SERVICES LIST

Exome-Seq

Efficient analysis of functional playersv.

Quality and Flexibility:

- EMQN certified pipeline
- Benchmarked with GIAB
- Whole and clinical exome
- Addition of extra targets

Supported Analyses:

- Variants functional annotation
- Trio analysis
- Tumor-normal matched pairs
- Mitochondrial variants
- SV and CNV detection

Access to the variant interpretation platform that combines artificial intelligence with ACMG-AMP guidelines for the accurate prioritization. An extremely intuitive interface allows genetic experts to focus on the smallest interesting set of variants.

CRISPR/Cas9-targeted enrichment and long-read sequencing

Accurate sequence analysis of complex genomic regions that cannot be explored with standard short-read NGS technologies.

- Target any genomic region regardless of sequence content
- Sequence through long repeats and low complexity regions
- Count simple repeats expansions
- PCR-free enrichment of 2 to 20 kb regions
- Even coverage

Enrichment without amplification preserves native DNA molecules for sequencing, allowing for the direct detection and characterization of epigenetic signatures.

Whole Genome Sequencing

All genetic tests in one.

- Capture both large and small variants
- Survey non-coding regions and clinically relevant SVs and CNAs
- PCR-free library preparation
- *De novo* assembly of personal genomes

ADVANCED BIOINFORMATICS ANALYSIS

- Comparative genomic analysis - characterize differences and similarities between genomes from the same or different species.
- Identification of somatic and germline structural variants
- Event-specific insertion analysis

QC during the sample processing ensures the delivery of the highest quality data available

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