DNA
Service Specification Sheet

Genome Scan

Long Read | Methylation Arrays | Targeted | WES | WGS

Whole Genome Sequencing (WGS)

The most comprehensive sequencing analysis with the widest coverage

Whole Genome Sequencing (WGS) provides the most comprehensive genetic blueprint of an organism. WGS is often conducted in exploratory analysis, for example in the investigation of rare diseases of unknown causes. GenomeScan offers an accredited (ISO/IEC 17025) WGS service across a multitude of diagnostics, therapeutics and research applications. WGS not only provides unique insights into disease mechanisms, but it also generates genome wide information opposed to Whole Exome Sequencing (WES) generating data for only 1-2% of the genome. It offers the ability to identify copy number variations, insertions-deletions (indels), rearrangements and other structural variations. It helps you make an educated decision for the next step of your research project and offers you the advantage of reanalyzing and reinterpreting your data at any time.
By using unique dual-indexed sequencing adaptors, we are able to analyze hundreds of samples in parallel delivering the least bias and highest reproducibility, making WGS a very powerful and cost-effective tool for genetic profiling.

Input material

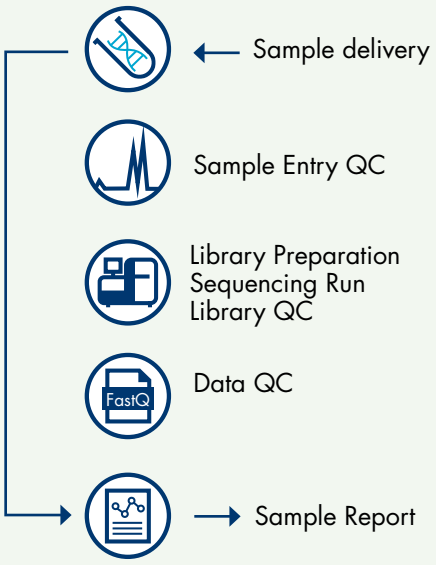
Isolated genomic DNA
- Required input:
  - without PCR ≥ 25 ng / sample*
  - with PCR ≥ 1 ng / sample
- Minimum volume: 20 µl / sample
- Quality: High Molecular Weight DNA
- Optional: FFPE repair available

Sequencing on Illumina NovaSeq (PE 150)
- PCR free library preparation (≥ 25 ng)
- Unique Molecular Identifiers for samples with PCR
- Unique dual indexed sequencing adaptors

Deliverables
- FastQ files
- Quality score Q30 of ≥ 80%
- Optional data analysis with comprehensive report

*Higher input is required (> 100 ng) for deep sequencing

NGS Laboratory workflow



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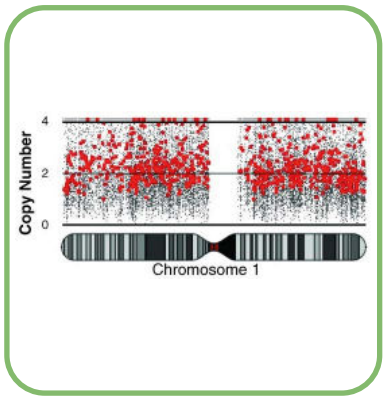
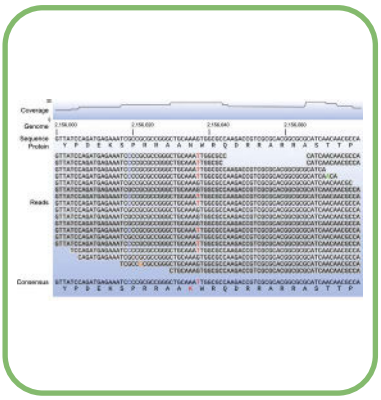
ISO/IEC 17025
L518 By RvA



Mutation map on chromosome



Genetic variants detection

De novo assembly

Committed to your project

Data quality guarantee
The purpose of data QC is to validate the precision and accuracy of your results. It helps to reduce the time you spend on sample verification.

ISO 15189 and ISO/IEC 17025 accreditation
The company strives for excellence and is committed to driving up standards in the field of NGS services. For our customers, this means consistency in our level of quality, service and support.

Reporting/Publication ready results
We deliver comprehensive, consistent and transparent NGS information. Furthermore, we offer different visualization options to make your results ready for presentation and/or publication.

Data analysis options

To understand the effect of genomic variations, the next step in your sequencing project is the analysis of millions of high-quality reads produced during the sequencing runs. At GenomeScan, we have a team of dedicated bioinformaticians that combine bioinformatics and statistical approaches with high-performance computing to assist you with the interpretation of your data. All information is manually reviewed by experts to comply with our high-quality standards and generate results you can trust.
For optimal interpretation of the data, our workflow covers preprocessing steps that include data trimming and alignment to reference sequences, followed by variant calling using golden standards. Afterwards we annotate variants for functional and disease relevance using multiple databases. Our data analysis report provides several visualization possibilities (see figures).

Biological Insights

The biological insights that can be inferred from your data include:

- All variant types reported: Single nucleotide polymorphisms (SNPs), indels, multi-nucleotide polymorphism (MNPs), and copy number variants (CNVs)
- Genomic rearrangements
- *de novo* genome assembly and refinement
- Annotation of variants

Custom solutions

At GenomeScan, we continuously improve our services and validate new solutions. Take advantage of our pilot program in which bioinformatics pipelines can be tailored to your specific needs, allowing more in-depth mining of your dataset. In addition to that, we offer various customizable publication grade visualization options to present your results.

About GenomeScan

As an ISO-accredited leading Dutch Next Generation Sequencing service provider, GenomeScan develops customizable NGS solutions for pharmaceutical and biotech companies, healthcare providers and academic institutions. By providing state-of-the-art tools to analyze genetic disorders fast, affordably, and effectively, GenomeScan fosters innovation through partnership with medical centers and research laboratories.

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