Product Specification Sheet

Genome Scan

Chip | Longreads | Methyl arrays | Targeted | **WES** | WGBS | WGS

Whole Exome Sequencing (WES)

Highly reliable insights into protein-coding regions

Since most known inherited disease causing mutations are found in exons, Whole Exome Sequencing (WES) is a very efficient diagnostic screening method. This method is used for facilitating the discovery of common and rare mutations, as well as to identify other alterations in genes, such as small deletions and insertions. WES covers ~20.000 genes and is offered as an ISO/IEC 17025 accredited service at GenomeScan. The Agilent SureSelect library preparation (capture-based technique) generates high quality data for diagnostic purposes. WES provides higher read depth (>100x) in a cost-effective manner and can be completed in one week. This method generates a more concise data set that allows faster data analysis.

GenomeScan developed a validated protocol for WES on FFPE material that represents an important and innovative alternative for possible applications in a clinical environment.

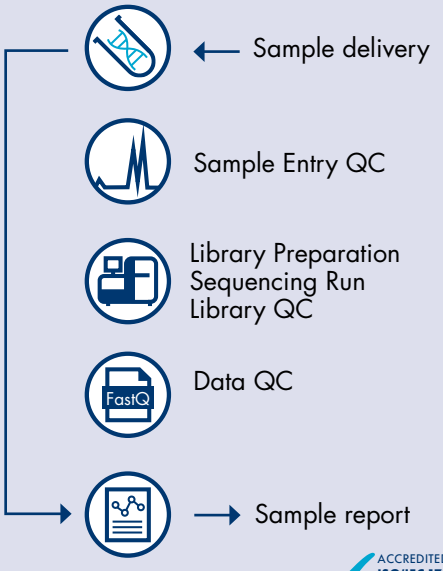
Input material

Purified DNA
- Validated input: ≥ 20 ng / sample
- Minimum volume: 15 µl / sample
- Quality: Column or bead purified DNA

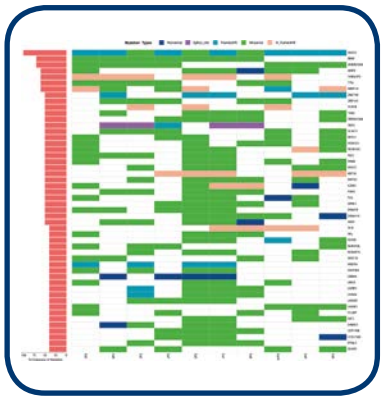
Sequencing on NovaSeq 6000
- Standard All Exon V7 + CNV backbone bait library
- Unique dual indexed sequencing adaptors
- Standard read depth 40M reads / sample

Deliverables
- TAT: 2 weeks after successful sample QC
- FastQ files via secured electronic transfer
- Quality score Q30 of ≥ 80% for PE150 reads
- Optional data analysis with comprehensive report resulting in BAM and VCF files

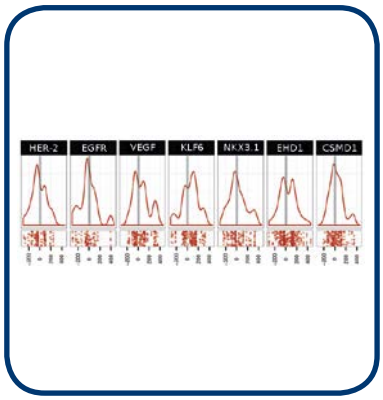
NGS Laboratory workflow



Variant call and annotations



Mutation enrichment per gene



Coverage for target regions

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 WES a very reliable and cost-effective tool for genetic profiling in both diagnostics and research settings.

Committed to your project

Data quality guarantee:
Sequence quality control is an essential tool in our workflow. We track, identify and exclude potential errors that could impact the interpretation of your results.

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 data:
We deliver comprehensive, consistent and transparent NGS information, Furthermore, we offer different visualization options to make your results ready for reporting.

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 provide you with a fast biological 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).

Applications
Types of projects and applications where WES can particularly be powerful include:

- Drug trials and pharmacogenetic studies
- Analysis of large-scale cohorts
- Analysis of mutation enrichments in genes
- Tracing Mendelian disorders in families
- Personalized medical interventions

Customized Pipelines

If necessary, we can set-up a custom analysis plan with you. Bioinformatic pipelines can be tailored to your research and/or clinical needs allowing more in-depth mining of your dataset when necessary. We generate reports that optimally address your research and/or clinical question.

About GenomeScan

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

genomescan.nl

+31 71 5681050

info@genomescan.nl