Service Specification Sheet

Genome Scan

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

Whole Genome Bisulfite Sequencing (WGBS)

Golden standard of methylation profiling

Methylation of cytosines in DNA molecules is a conserved epigenetic modification from bacteria to eukaryotes. It is a regulator of transcriptional activity and plays an important role in embryonic development, genomic imprinting, transposon silencing, X-chromosome inactivation and pluripotent cell differentiation. Differential methylation patterns are also indicated in disease states, making methylation profiling a useful tool for uncovering disease mechanisms, predictive biomarkers and drug targets. Bisulfite sequencing has been considered the golden standard of DNA methylation analysis and combining it with NGS enables global genomic profiling of cytosine methylation in an effective manner.
At GenomeScan, we also use unique dual indexed sequencing adaptors with UMIs for results of the highest reliability, which then become easily accessible through our web-based user portal.

Input material

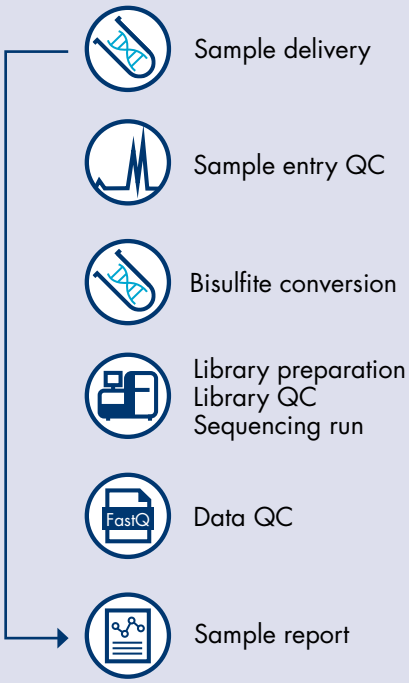
Isolated genomic DNA

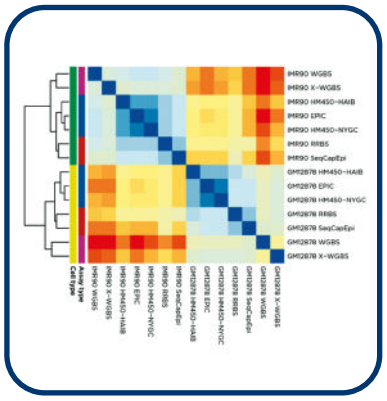
Isolated sample requirements
- Preferred minimal input: ≥100 ng / sample
- Minimal volume of 20 µl / sample
- HMW genomic DNA, column or bead purified

Sequencing on Illumina NovaSeq (PE 150)
- Standard read depth of x30
- Unique dual indexing combined with unique molecular identifiers (UMIs)

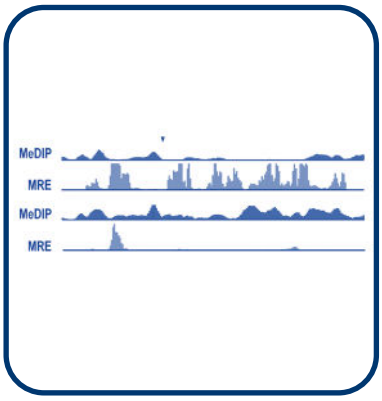
Deliverables
- FastQ files
- Quality score (Q30) ≥ 80%
- Optional data analysis with comprehensive report

NGS Laboratory workflow

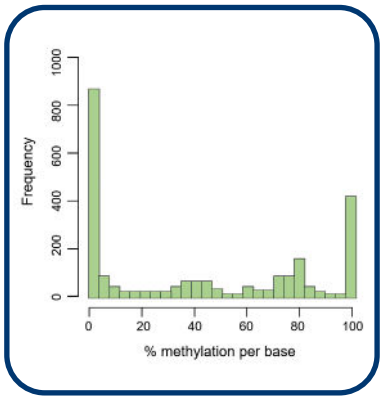




Concordance of DNA methylation



CpG island methylation coverage



Methylation distribution

Committed to your project

Data quality guarantee:
Sequence quality control is an essential step in our workflow. We perform several controls during the preparation and analysis of WGBS datasets to guarantee performance and quality of sequencing outputs.

Single nucleotide resolution:
We have developed computational models to provide biological interpretation of your results. Our service makes genome wide DNA methylation mapping of a large number of samples possible at a high resolution.

Publication ready results:
We deliver comprehensive, consistent and transparent NGS information. Bioinformatics methods facilitate visualization of your DNA methylation data, which can directly be used in your presentations and manuscripts.

Data analysis options

Whole genome sequencing of bisulfite treated DNA samples results in a large set of data. Interpreting these results for answering your particular research or clinical question calls for expert input. At GenomeScan, we have a dedicated bioinformatics team with extensive experience in genomic data analysis and interpretation. They combine bioinformatics with high-performance computing to provide you with comprehensive and ready-to-publish results.

For reliable interpretation of your data, our workflows cover optimized quality check, preprocessing, alignment, and methylation level determination steps at each nucleotide, as well as a differential analysis of cytosine methylation between samples. From raw sequencing data to statistically processed results, our data analysis report makes your results easy to interpret and ready to present or publish.

Applications

Projects that benefit from whole genome DNA methylation profiling include:

- Non-human DNA methylation profiling
- Study of clinical cohorts
- Patient stratification
- Study of age-related pathologies, such as cancer, osteoarthritis, and neurodegeneration
- Genetic disease marker identification
- Identification of possible drug targets
- Personalized medicine development

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 topresent 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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