Service Specification Sheet

Genome Scan

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

Array Based Genome Wide Methylation Analysis

Profiling methylation of CpG islands and more

Epigenetic processes play important roles in health and diseases. DNA methylation is a key component of gene regulation. Mapping DNA methylation sites in clinical samples can potentially identify genes linked to specific disease mechanisms, drug targets or predictive biomarkers for disease diagnostics. Interpreting DNA methylation data can also facilitate quality control of cultured cells as cell states are reflected in their DNA methylation patterns.

GenomeScan works with Illumina's high density Infinium MethylationEPIC BeadChip, which offers a comprehensive genome wide coverage not only of CpG islands but also other CpG and non-CpG methylated sites identified in human stem cells, a variety of promoter and enhancer sites, and DNase hypersensitive sites. The automated workflow results in an unprecedented high concordance between arrays and the results are accessible through our web-based user portal.

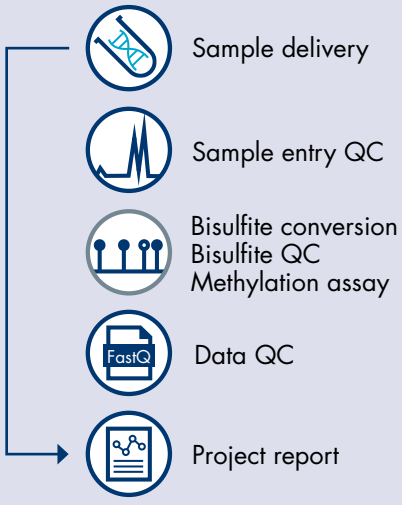
Input material

Isolated genomic DNA
- Validated input: ≥ 750 ng / sample
- (based on fluorescent measurement)
- Minimum volume: 20 µl / sample
- Quality: Column or bead purified DNA

MethylationEPIC BeadChip
- ±935K methylation sites
- Includes 99% of RefSeq genes
- Covers 95% of CpG islands

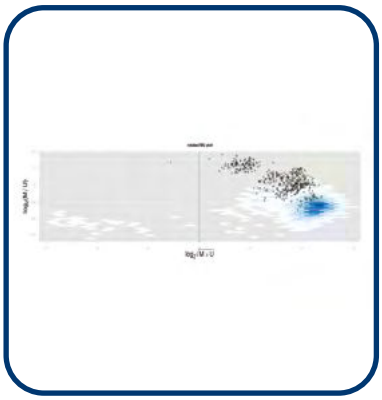
Deliverables
- TAT: 8 weeks after successful sample QC
- iDAT files via secured electronic transfer
- Comprehensive QC report
- Optional data analysis with comprehensive report

NGS Laboratory workflow

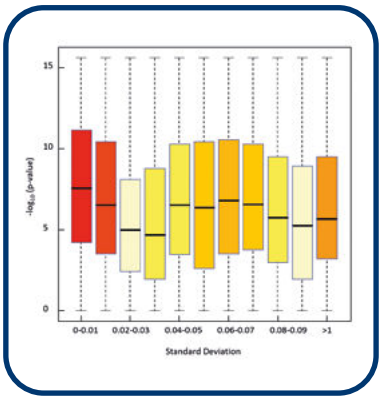


Control metric	Type of control	Result
Staining	Sample-independent	QC Passed
Hybridization	Sample-independent	QC Passed
Target Removal	Sample-independent	QC Passed
Extension	Sample-independent	QC Passed
Bisulfite Conversion I	Sample-dependent	QC Passed
Bisulfite Conversion II	Sample-dependent	QC Passed
Specificity I	Sample-dependent	QC Passed
Specificity II	Sample-dependent	QC Passed
Negative	Sample-dependent	not included
Non-polymorphic	Sample-dependent	QC Passed

System control for EPIC array



Bisulfite conversion control



DNA methylation variability

Committed to your project

Data quality guarantee:
By adding a positive control sample to your set of samples, the expected high data quality can be checked by us before sending you the results.

Interpretation of methylation data:
We have developed computational models to process, analyze and interpret large-scale DNA methylation sets. 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 information. Bioinformatic methods for visualization of your DNA methylation data facilitate results presentation and help prepare your manuscript.

Data analysis options

Not a single method of DNA methylation analysis will be appropriate for answering every research question. At GenomeScan, we have access to a 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 optimal interpretation of the data, our workflow covers preprocessing steps that include import of the raw methylation data, quality control, filtration and normalization, data exploration and analysis. It is optimized to process multiple samples in parallel.
One important aspect of exploring results of an analysis is visualization. From raw sequencing data to statistically processed results of differentially methylated genes, our data analysis report provides several visualization possibilities (see figures above).

Applications

Projects that benefit from DNA methylation profiling by Infinium arrays include:

- 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 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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