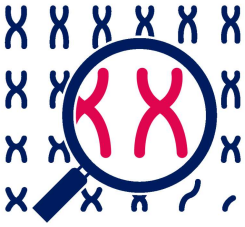
Whole Genome Sequencing

GENERAL *DETAIL* *REPORT*

Do you need to sequence the complete genomic information of your samples? Macrogen Europe's fast, high-quality WGS services provide the most comprehensive tool for detecting genetic abnormalities such as single nucleotide polymorphisms (SNPs), indels, copy number variation (CNV) and structural variants (SVs).



WGS analyses an organism's entire genome, as opposed to *whole exome sequencing* and *targeted sequencing*, which studies specific areas. **A single test identifies all variations in the genome** such as single-nucleotide variants, insertion, deletion, copy number variants, and translocations. This approach covers both the coding and non-coding parts of the genome. For this reason, **WGS is the sequencing option of choice to study factors that affect the entire genome and to analyze noncoding DNA.**

Emerging technologies provide the better resolution of challenging regions containing highly variable and repetitive elements, supported by a readily available long-read sequencing platform and proficient library preparation kit selection from Macrogen, thereby speeding the adoption of this testing into routines.

APPLICATIONS

This is particularly beneficial when unexplained conditions are to be discovered in vital genetic diagnosis and treatment. WGS is essential in a wide range of scientific fields, both fundamental and applied. It can **identify genetic variations across the genome,** which is why it is commonly used in agriculture and evolutionary biology and in clinical and pharmaceutical applications such as **infectious diseases, immunology, cancer research, various inherited conditions and drug development.**

SHORT-READ VS LONG-READ SEQUENCING

>99.9% accuracy sequencing data for large batches and low budget per sample

VS

Continuous long sequence data with improve accesibility to AT/GC-rich regions

Short-read sequencing

The genome is randomly fragmented in fragments of 100-600bp

Libraries are sequenced to produce reads up to 300bp

NovaSeq X | MiSeq | NextSeq | NovaSeq 6000

Long-read sequencing

The genome is randomly fragmented in fragments of thousand of base pairs

Libraries are sequenced to produce reads up to 300Kbp

Sequel IIe | Revio | GridION

Combine short-read and long-read sequencing to fill gaps or correct errors during genome assembly

DE NOVO SEQUENCING VS RESEQUENCING

De novo sequencing offers a way to uncover genome information about microorganisms, animals and plants when no reference genome is available. The analysis works by assembling long parallel phases of genetic material without referencing an existing nucleotide sequence. This type of WGS is highly accurate and can replicate or complete a blueprint for complex or polyploid genomes.

Resequencing a genome involves comparing newly sequenced DNA to a previously created reference genome. It is a highly efficient, fast and cost-effective way of analyzing the genomes of commonly studied organisms. Whole genome resequencing can scan or test for specific genetic variations. Any differences between the sample DNA and the reference could be an indication of a new variant or a potential disorder in a clinical setting.

What is mappable coverage? What coverage do you need?

Understanding next-generation-sequencing can be challenging. Knowing how much data your project needs is essential. Read and learn with Macrogen Europe.

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