WHOLE GENOME SEQUENCING

A comprehensive look at an individual's entire genome.

- To identify all variation in the genome
- Coding and non-coding genome
- To discover unexplained conditions in vital genetic diagnosis and treatment

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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

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

VS

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

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

TIP

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

ZOOM

LIBRARY KITS AND CRITERIA

SHORT READ SEQUENCING

Input	FFPE	Shearing	PCR Free	Library kit
1ng	No	Enzymatic	No	***Nextera XT DNA***
100ng	Yes / No	Mechanical	No	***TruSeq Nano***
200ng	Yes	Enzymatic	Yes / No	***Watchmaker DNA***
500ng	No	Enzimatic	Yes / No	***Illumina DNA***
1000ng	No	Mechanical	No	***TruSeq DNA***

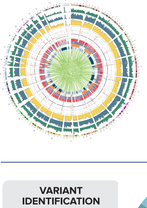
We advise sending samples in a volume ranging from 25 to 50 µl to prevent them from drying out during shipment

LONG READ SEQUENCING

Input	(HMW)	Multiplexing	Platform	Library kit
4µg	Average 15Kbp	Yes (96plex)	***Sequel IIe Revio***	***SMRTbell Microbial***
6µg	Average 30Kbp	Yes (16 plex)	***PromethION***	***ONT LSK114***
7µg	Average 20Kbp	Yes (96 plex)	***Sequel IIe Revio***	***SMARTbell HiFi***

The kits mentioned above are the most frequently used. For more kits please, contact our team.

BIOINFORMATICS



MAPPING

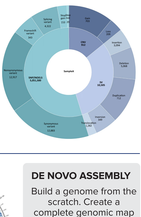
Align sequencing reads to a reference genome to identify their origin. It enables the study of variations and genome structure

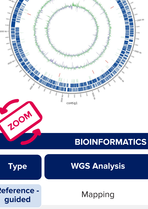
ANNOTATION

Identify functional genes and regulatory regions to understand their role and interactions

VARIANT IDENTIFICATION

To identify difference in DNA sequences between individuals (or reference genome), allowing for the identification of genetic variations associated with traits or diseases





DE NOVO ASSEMBLY

Build a genome from the scratch. Create a complete genomic map without genome reference

ANNOTATION

Identify functional genes and regulatory regions to understand their role and interactions

ZOOM

BIOINFORMATICS OVERVIEW

Type	WGS Analysis	Short-read	Long-read
Reference - guided	Mapping	●	●
	Variant calling (SNP/Indel)	●	●
	Copy number variant identif.	●	●
	Structural variant identif.	●	●
	Insertion site prediction	●	
	CpG Analysis		●
	Joint Genotyping	●	
	Family Analysis	●	
	Somatic Analysis	●	
	Telomere Analysis	●	
	sgRNA Analysis	●	
	Mitochondrial DNA Heteroplasmy	●	
	Microbial Classification	●	
De novo	De Novo Assembly and Validation	●	●
	Genome Survey	●	●
	Gene Prediction and Annotation	●	●
	Whole Genome Alignment	●	●
	Phylogenetic Analysis	●	

Macrogen Europe's analysis is not limited to the services listed above. The visualization, pangenome analysis, and the other analysis are serviced upon the discussion.

THE HOUSE OF SEQUENCING

Humanizing Genomics macrogen EUROPE

POWERED BY

illumina · PacBio · NANOPORE · WATCHMAKER GENOMICS · ThermoFisher SCIENTIFIC · TWIST BIOSCIENCE · Singleron · Agilent Technologies · 10X GENOMICS