WHOLE EXOME SEQUENCING

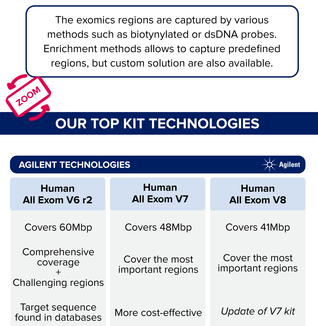
A powerful application that focus on protein-coding regions

The 85% of known disease-related genetic mutations are found in exomic regions

WES demands less throughput than WGS, making it more cost-effective

The method of choice for many researchers and clinicians

© 2024 Macrogen Europe - Whole Exome v1



The exomics regions are captured by various methods such as biotynylated or dsDNA probes. Enrichment methods allows to capture predefined regions, but custom solution are also available.

ZOOM

OUR TOP KIT TECHNOLOGIES

AGILENT TECHNOLOGIES

Human All Exom V6 r2	Human All Exom V7	Human All Exom V8
Covers 60Mbp	Covers 48Mbp	Covers 41Mbp
Comprehensive coverage + Challenging regions	Cover the most important regions	Cover the most important regions
Target sequence found in databases	More cost-effective	*Update of V7 kit*
0.5 µg	0.5 µg	0.5 µg

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

TWIST BIOSCIENCE

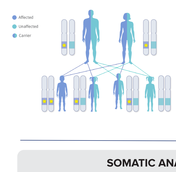
Core Exome	Comprehensive Exome	TWIST 2.0 Exome
Covers 33Mbp	Covers 41Mbp	Covers 36.5Mbp
Specially targetted to CCDS* regions	Target CCDS + RefSeq and GenCode databases	Target CCDS + RefSeq and GenCode databases + Clinvar and other relevant databases
50ng	50 ng	50 ng

*Consensus Coding Sequence

For TWIST, a min 50ng is required for input. However, Macrogen Europe recommends to send 200ng for optimal results.

The kits mentioned above are the most frequently used. For more kits such as UTRs, mtDNA, Cosmic probes, please, contact our team

BIOINFORMATICS



FAMILY ANALISIS

Identify inherited traits or diseases by comparing familial genomes, aiding in personalized medicine and understanding inheritance patterns

SOMATIC ANALYSIS

Somatic analysis in sequencing identifies mutations in non-reproductive cells that occur during a person's lifetime. This is key in cancer research for detecting tumor-specific mutations and guiding targeted therapies.

ZOOM

BIOINFORMATICS OVERVIEW

Type	Analysis	Description
Standard	**Variant Calling** (SNP/Indel)	Identifying genetic differences in sequenced DNA compared to a reference genome, essential for understanding genetic variation and disease.
	Variant Annotation	Adds information to genetic variations, aiding interpretation of genomic data for understanding their functional significance in health and disease
Advanced	**CNV** (Copy Number Variation)	Elucidates changes in DNA copy numbers, helping understand their role in genetic variation and disease, aiding interpretation of genomic data.
	Cancer Analysis	Examines genomic changes in cancer cells to understand the disease, identify treatment targets, and personalize therapies.
	Population Analysis	tudies genetic diversity to understand evolution, traits distribution, and disease risk, informing medical and public health efforts.

The Advanced Bioinformatics services listed above are not available for custom target sequencing. Please, contact our team about extra BI services

THE HOUSE OF SEQUENCING

Humanizing Genomics macrogen EUROPE

POWERED BY

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