GENEQUALITY®
Whole Exome Sequencing

GENEQUALITY® Whole Exome Sequencing is a high-throughput Next-Generation Sequencing (NGS) assay designed to capture and sequence the protein-coding exonic regions of the human genome (gDNA) and mitochondrial DNA (mtDNA).

The GENEQUALITY® Whole Exome sequencing features a highly sensitive kit for nucleic acid library preparation and enzymatic fragmentation, used to create sequencing libraries through a target capture system.

For *in vitro* diagnostic use

ANALITICA
ADVANCED BIOMEDICINE

GENEQUALITY®
Whole Exome Sequencing

Whole Exome Sequencing (WES) is a high-throughput Next-Generation Sequencing (NGS) method that captures and sequences the protein-coding regions (exons) of the human genome, covering approximately 20,000 genes[1,2].

Although these exonic regions represent only 1% of the genome, they contain up to 85% of pathogenic mutations[3].

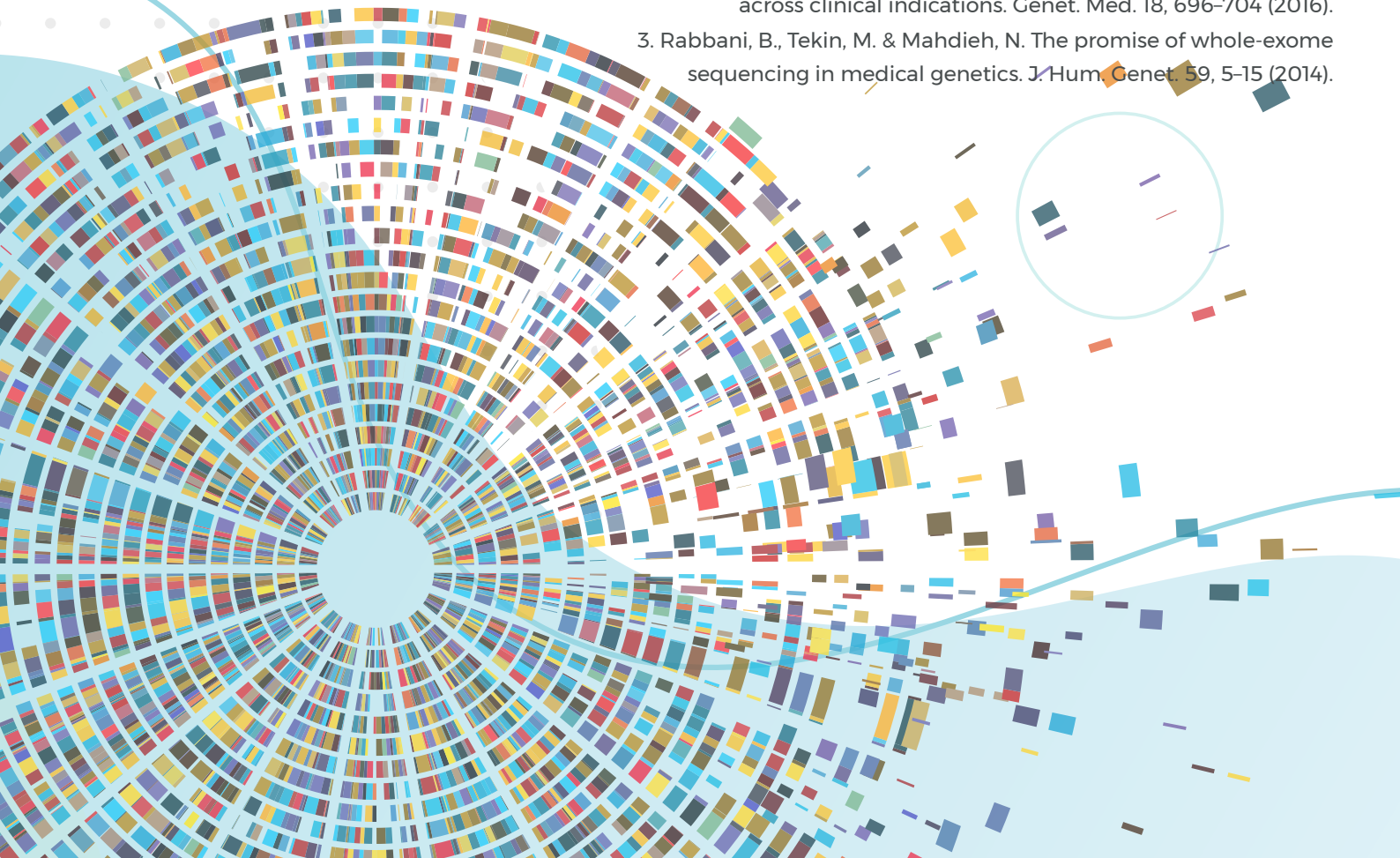
The **GENEQUALITY® Whole Exome Sequencing** includes a highly sensitive nucleic acid library preparation and enzymatic fragmentation kit used for the preparation of sequencing libraries from high-quality genomic DNA (gDNA) and and mitochondrial DNA (mtDNA) isolated from human cells, such as whole blood. The kit utilizes a target capture system to produce target enriched libraries compatible with Illumina sequencing instrumentation.

The **GENEQUALITY® Whole Exome Sequencing** covers 19,441 genes and the entire mitochondrial genome, enabling an exome-wide investigation. Probe design is highly optimized to guarantee a high on-target reads percentage and coverage uniformity even in GC-rich regions.

1. Yang, Y. et al. Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. N. Engl. J. Med. 369, 1502–1511 (2013).

2. Retterer, K. et al. Clinical application of whole-exome sequencing across clinical indications. Genet. Med. 18, 696–704 (2016).

3. Rabbani, B., Tekin, M. & Mahdieh, N. The promise of whole-exome sequencing in medical genetics. J. Hum. Genet. 59, 5–15 (2014).



GENEQUALITY®
Whole Exome Sequencing Workflow

Blood collection

Library preparation

Sequencing

Interpretation & reporting

Clinical managment

ENZIMATIC FRAGMENTATION

5-10 min hands on time
55 min incubation

GENEQUALITY Library Prep ref. No. 04-N01-6M

ADAPTERS LIGATION

5-10 min hands on time
15 min incubation

GENEQUALITY Unique Dual Indexes ref. No.04-N03-6M

PURIFICATION

20 min hands on time

GENEQUALITY Purification kit ref. No.04-N02-6M

LIBRARY AMPLIFICATION

5-10 min hands on time
15 min incubation

GENEQUALITY Library Prep ref. No. 04-N01-6M

PURIFICATION

20 min hands on time

GENEQUALITY Purification kit ref. No.04-N02-6M

QC STEP 1

Stopping point

pre-PCR area

POOL CREATION

Library 1
Library 2
Library 3

5 min hands on time

POOL CONCENTRATION

Library 1
Library 3
Library 2
Library 3
Library 1
Library 2

20 min hands on time

GENEQUALITY Whole Exome Sequencing ref. No. 04-NWE-6M

Overnight

HYBRIDIZATION

5-10 min hands on time
16 hrs incubation

GENEQUALITY Whole Exome Sequencing ref. No. 04-NWE-6M

CAPTURE WITH STRPTAVIDIN BEADS

1,5 hrs hands on time

LIBRARIES AMPLIFICATION

5-10 min hands on time
15 min incubation

GENEQUALITY Whole Exome Sequencing ref. No. 04-NWE-6M

PURIFICATION

20 min hands on time

GENEQUALITY Purification kit ref. No.04-N02-6M

QC STEP 2

Stopping point

SEQUENCING

ANALYSIS/REPORT

post-PCR area

Sequencer Loading Scheme

Sequencer	Reagent system	Read lenght	Sample / Flow cell	Total Flow cell output
NovaSeq® 6000	S2 Reagent Kit	2x150	100	1000-1250 Gb
	S4 Reagent Kit	2x150	250	2400-3000 Gb
NextSeq® 2000	P3 XLEAP-SBS Reagents	2x150	36	360 Gb
	P4 XLEAP-SBS Reagents	2x150	54	540 Gb
NextSeq® 500/550	High-output flow cell	2x150	12	100-120 Gb

Estimated number of samples that should be loaded per flow cell to reach a mean target coverage of ~100X.

Databases and % covered by GENEQUALITY® WES (GQ WES)

HGMD
version 2024.1

Tot	%	DM/DM?	% Pat
474,280	96.43	465,618	97.00

Tot Total Variants
% % variants covered by GQ WES
DM Disease-causing Mutation covered by GQ WES
DM? Likely Disease-causing Mutation covered by GQ WES
% Pat % pathogenic variants covered by GQ WES

Human Gene Mutation Database (HGMD®) is a comprehensive collection of germline mutations in nuclear genes that underlie, or are associated with, human inherited disease.

ClinVar
version 2024/01/04

Tot	%	Path/LPath	% Pat
2,347,099	87.58	232,675	98.15

Tot Total Variants
% % variants covered by GQ WES
Path Pathogenic Variants covered by GQ WES
LPath Likely Pathogenic Variants covered by GQ WES
% Pat % pathogenic variants covered by GQ WES

ClinVar is a freely accessible, public archive of reports of human variations classified for diseases and drug responses, with supporting evidence.

OMIM
version 2024/04/02

Genes	% genes	Positions	% positions
4,936	99.52	459,152,651	3.23

Genes OMIM genes
% genes % OMIM genes with associated phenotypes covered by GQ WES
Positions OMIM positions with associated phenotypes covered by GQ WES
% positions % OMIM positions with associated phenotypes covered by GQ WES

Online Mendelian Inheritance in Man (OMIM) is a continuously updated catalog of human genes and genetic disorders and traits, with a particular focus on the gene-phenotype relationship.

Uniformity

GENEQUALITY® WES offers excellent uniformity in sequencing coverage across regions with both low and high GC content.

Key metrics (clinical samples)

Key metrics	gDNA	mtDNA
Uniformity of coverage (Pct > 0.2*mean)	97.8%	99.9%
On-target bases	89.6%	90.8%
Detected Variants	SNVs Indels CNVs	
Average % of target region with depth >10x	99.7%	100%
Average % of target region with depth >20x	99.6%	100%
Average % of target region with depth >30x	99.0%	100%
Sensitivity (all variants)	95.81%	
Specificity (all variants)	99.99%	

GENEQUALITY® Library Prep, **GENEQUALITY® Purification Kit** and **GENEQUALITY® Unique Dual Indexes** are *in vitro* diagnostic devices used together by healthcare professionals to prepare libraries from high-quality genomic DNA for Next-Generation Sequencing (NGS) applications. These devices include reagents for enzymatic fragmentation, library preparation, purification, and universal adapters.

GENEQUALITY® Whole Exome Sequencing is another *in vitro* diagnostic device for producing exome and mitochondrial genome-enriched libraries for NGS-based examinations.

PRODUCT CHARACTERISTICS:

- Input sample: **human whole blood**
- DNA input requirement: **10 to 500ng (recommended 100ng)**
- Validated sequencing platform: **Illumina (NovaSeq6000, NextSeq500/550)**
- The panel size is approximately **42 Mb (19,441 genes covered)** and includes **mitochondrial genome**
- The kit is **CE-IVD** according to **Regulation EU 2017/746** (**IVDR**)
- The kit detects: **CNVs, InDels, SNPs, SNV**

Next-Generation Sequencing

EU-IVDR

ORDERING INFORMATION:

PRODUCT	CODE	FORMAT
GENEQUALITY® Library Prep	04-N01-6M	96 tests
GENEQUALITY® Purification kit	04-N02-6M	96 tests
GENEQUALITY® Unique Dual Indexes	04-N03-6M	96 tests
GENEQUALITY® Whole Exome Sequencing	04-NWE-6M	8x12 tests

AB ANALITICA ADVANCED BIOMEDICINE

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