




AviSeq<sup>TM</sup>  
Genomic DX




# NEXT GENERATION SEQUENCING LIBRARY PREPARATION KITS

[www.avcna.net](http://www.avcna.net)







# INTRODUCING AVISEQ, YOUR NEXT-GENERATION SOLUTION FOR LIBRARY PREPARATION



**Stable ready-to-use reagents**




**Wide range of human diseases target**




**Highly accurate detection**



**Compatible with multiple NGS platforms**



**Minimum DNA sample volume required**



**Simple and fast workflow**



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# AviSeq™ Wholesome

AviSeq™ Wholesome is a kit for the sequencing of the coding regions of the genome focusing the resources on the genes most likely to affect phenotype in subjects suffering from conditions not attributable to known pathologies, in subjects affected by genetically heterogeneous diseases and complex syndromic pictures.

AviSeq™ Wholesome allow the coverage of greater than 99% of protein coding genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues samples.

AviSeq™ Wholesome kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina.



## SOLUTION

- Comprehensive, accurate analysis of human whole exome regions.
- Based on recent database releases.
- Coverage uniformity and sequencing efficiency.



## TECHNOLOGY

The AviSeq™ Wholesome kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.

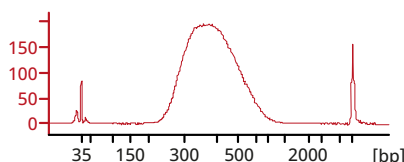


Figure 1. AviSeq™ Wholesome library Bioanalyzer profile.

## Sample per Run

Instrument	300x	100x	50x
NextSeq 550 Mid-Output Kit	4	8	16
NextSeq 550 High-Output Kit	8	24	48
NovaSeq 6000 SP	16	48	96

\*the estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ Wholesome (16 test)	AVG108016
AviSeq™ Wholesome (96 test)	AVG108096
UDI Primers Set A (96 test)	AVG406
UDI Primers Set B (96 test)	AVG407
UDI Primers Set C (96 test)	AVG408
UDI Primers Set D (96 test)	AVG409
UDI Primers Set 16 (16 test)	AVG410



# AviSeq™ BRCA

Hereditary variants profiling in Breast and Ovary Cancer



## PANEL

AviSeq™ BRCA is a kit for the identification of both of germline mutations in BRCA 1 and BRCA 2 genes.



## SOLUTION

AviSeq™ BRCA is a kit for the analysis of the BRCA1 and BRCA2 genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from cancer tissues (fresh, frozen or FFPE) or body tissues (blood or other).

AviSeq™ BRCA kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## WORKFLOW

The AviSeq™ BRCA kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.

Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing.

Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

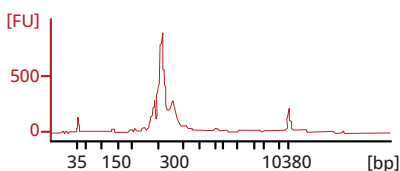


Figure 1. AviSeq™ BRCA library Bioanalyzer profile.

Sample per Run	
Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	18
MiSeq Nano Kit v2 (500-cycles)	18
MiSeq Micro Kit v2 (300-cycles)	80
MiSeq Kit v2 (300-cycles)	300
MiSeq Kit v2 (500-cycles)	300
MiniSeq Mid Output Kit (300-cycles)	160
iSeq 100 i1 kit (300-cycles) NextSeq	80
Ion 314™ Chip	3
Ion 316™ Chip	14
Ion 318™ Chip/Ion 520™ Chip Ion 530™ Chip	28
	>96

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

Ordering Information	
Test Per Kit	Kit Reference No.
AviSeq™ BRCA (16 test)	AVG104016
AviSeq™ BRCA (96 test)	AVG104096
<i>For Illumina instrument</i>	
AviSeq™ Index Set 16 (16 test)	AVG404
AviSeq™ Index Set 96 (96 test)	AVG411
AviSeq™ Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
Barcode Set 1-16 (96 test)	AVG502
Barcode Set 17-32 (96 test)	AVG503

Table 1. List of target regions in AviSeq™ BRCA

BRCA1	BRCA2
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# AviSeq™ BRCA PLUS



## PANEL

AviSeq™ BRCA PLUS is a kit for the analysis of the BRCA1, BRCA2 and TP53 genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies.

The kit is validated for somatic (SNPs, indels) and germline analysis (SNPs, indels, CNVs) of DNA extracted from cancer tissues (fresh, frozen or FFPE) or body tissues (blood or other).

AviSeq™ BRCA PLUS kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## KEY FEATURES

- Genes targeted: BRCA1, BRCA2, TP53
- Application: somatic, germline analysis
- Number of pools: 3
- Panel size: 26 kb
- Input DNA: 20ng/reaction
- Coverage: full coding exons plus
- padding regions (>20bp)

## Sample per Run

Instrument	Sample per Run	
	Germline	Somatic
MiSeq Nano Kit v2 (300-cycles)	6	1
Nano Kit v2 (500-cycles)	6	1
Micro Kit v2 (300-cycles)	28	2
MiniSeq Mid Output Kit (300-cycles)	57	5
iSeq 100 il kit (300-cycles)	28	2
Ion 314™ Chip	4	0
Ion 316™ Chip	18	1
Ion 318™ Chip/Ion 520™ Chip Ion 530™ Chip	35	2
	96	9

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
<i>Aviseq™ BRCA PLUS (16 tests)</i>	AVG100016
<i>Aviseq™ BRCA PLUS (48 tests)</i>	AVG100048
<i>For Illumina instrument</i>	
Aviseq™ UDI Primers Set A (96 tests)	AVG406
Aviseq™ UDI Primers Set B (96 tests)	AVG407
Aviseq™ UDI Primers Set C (96 tests)	AVG408
Aviseq™ UDI Primers Set D (96 tests)	AVG409
Aviseq™ UDI Primers Set 16 (16 tests)	AVG410
<i>For Ion Torrent instrument</i>	
Aviseq™ Barcode Screen 1-16 (96 tests)	AVG900
Aviseq™ Barcode Screen 1-96 (96 tests)	AVG901



# AviSeq™ CARDIO

For the characterization of familial cardiovascular diseases



## PANEL

The AviSeq™ CARDIO kit is designed for the characterization of familial cardiovascular diseases. The panel summarizes together all genes described as related to inherited cardiomyopathies, rare diseases involving heart and hereditary aneurysmal diseases, following all the newest guidelines and recommendations (i.e. ClinGen). Both SNVs and CNVs can be identified in germline or somatic samples.



## TECHNOLOGY

The AviSeq™ CARDIO kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straight forward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.

## Sample per Run

Instrument	Sample per Run
MiSeq vi (300-cycle)	16
MiSeq Kit v2 (500-cycles)	16
MiSeq Kit v3 (600-cycles)	28
MiniSeq Mid Output Kit (300-cycles)	8
MiniSeq High Output Kit (300-cycles)	28
NextSeq 550 Mid-Output Kit	144

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ CARDIO (16 test)	AVG303016
AviSeq™ CARDIO (96 test)	AVG303096
<i>For Illumina instrument</i>	
AviSeq™ UDI Primers Set A (96 test)	AVG406
AviSeq™ UDI Primers Set B (96 test)	AVG407
AviSeq™ UDI Primers Set C (96 test)	AVG408
AviSeq™ UDI Primers Set D (96 test)	AVG409
AviSeq™ UDI Primers Set 16 (96 test)	AVG410

Table 1. List of target regions in AviSeq™ CARDIO

ABCC9	APOE	COL5A2	DTNA	GATAD1	KCND3	LDLRAP1	MT-TH	MYPN	PRKAG2	SCN5A	TAZ	TPM1
ABCG5	BAG3	COL5A1	EFEMP2	GCKR	KCNE1	LMF1	MT-TK	NEBL	PRKAR1A	SCO2	TBX20	TRDN
ABCG8	BGN	COX15	ELN	GJAS	KCNE2	LMNA	MT-TS1	NEXN	PRKGI	SDHA	TBX3	TRIM63
ACTA1	BRAF	CREB3L3	EMD	GLA	KCNE3	LOX	MT-TQ	NKX2-5	PSEN1	SEPN1	TBX5	TRPM4
ACTA2	CACNA1C	CRELD1	EYA4	GPDIL	KONH2	LPL	MURC	NODAL	PSEN2	SGCB	TCAP	TTN
ACTC1	CACNA2D1	CRYAB	FBN1	GPIHBP1	KCNJ2	LRRRC10	MYBPC3	NOTCH1	PTPN11	SGCD	TGFBR2	TTR
ACTN2	CACNB2	CSR3	FBN2	HADHA	KCNJ5	LTBP2	MYH11	NPPA	RAF1	SGCG	TGFBR3	TXNRD2
AKAP9	CALM1	CTF1	FHL1	HCN4	KCNJ8	MAP2K1	MYH6	NRAS	RANGRF	SHOC2	TGFBR1	VCL
ALMS1	CALR3	DES	FHL2	HFE	KCNQ1	MAP2K2	MYH7	OBSCN	RBM20	SLC25A4	TGFBR2	ZBTB17
ALPK3	CASQ2	DMO	FKRP	HRAS	KLF10	MAT2A	MYL2	PCSK9	RYR1	SLC2A10	TMEM43	2HX3
ANK2	CAV3	DNAJC19	FKTN	MSPB8	KRAS	MFAP5	MYL3	PDLIM3	RYR2	SMAD2	TMPO	ZIC3
ANKRD1	CBL	DOLK	FINA	ILK	LAMA2	MIB1	MYLK	PKP2	SALL4	SMAD3	TNNC1	
APOA4	CBS	DPP6	FUNC	JAG1	LAMA4	MT-TS2	MYLK2	PLEKHM2	SCN1B	SMAD4	TNNC2	
APOA5	CETP	DSC2	FOXE3	JPH2	LAMP2	MT-ND1	MYO6	PLN	SCN2B	SNTAL	TNNI3	
APOB	COL3A1	D562	FXN	JUP	LD83	MT-ND5	MYOM1	POLG1	SCN3B	S051	TNNI3K	
APOC2	COLSA1	DSP	GAA	KCNAS	LDLR	MT-ND6	MYOZ2	PRDM16	SCN4B	SREBF2	TNNT2	



# AviSeq™ CFTR

For the diagnosis of hereditary variants in Cystic Fibrosis



## PANEL

AviSeq™ CFTR is a CE-IVD kit for the molecular profiling of CFTR gene. Mutations in the CFTR gene cause the CFTR protein to malfunction or not produce, leading to a build-up of thick mucus, which in turn leads to persistent lung infections and multi-organ complications. More than 2200 causative mutations of Cystic Fibrosis have been described and with the AviSeq™ CFTR kit it is possible to detect all variants, known and unknown, with a single test. The importance of an NGS test is reflected in the timeliness of diagnosis in newborn screening, in the diagnosis of Cystic Fibrosis in asymptomatic patients, in the diagnosis of CFTR related diseases and in the possibility of directing the therapeutic path.



## SOLUTION

AviSeq™ CFTR is a kit for the analysis of the CFTR gene through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from body tissues (blood or others). AviSeq™ CFTR kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina.



## WORKFLOW

The AviSeq™ CFTR kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	18
MiSeq Nano Kit v2 (500-cycles)	18
MiSeq Micro Kit v2 (300-cycles)	80
MiSeq Kit v2 (300-cycles)	300
MiSeq Kit v2 (500-cycles)	300
MiniSeq Mid Output Kit (300-cycles)	160
iSeq 100 i1 kit (300-cycles)	80
Ion 314TM Chip	3
Ion 316TM Chip	14
Ion 318TM Chip/Ion 520TM Chip Ion	28
530TM Chip	>96

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ CFTR (16 test)	AVG300016
AviSeq™ CFTR (96 test)	AVG300096
<i>For Illumina instrument</i>	
AviSeq™ Index Set 16 (16 test)	AVG404
AviSeq™ Index Set 96 (96 test)	AVG411
AviSeq™ Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
AviSeq™ Barcode Set 1-16 (96 test)	AVG502
AviSeq™ Barcode Set 17-32 (96 test)	AVG503





# AviSeq™ CHOLESTEROL

For the genetic characterization of Familial Hypercholesterolemia



## PANEL

AviSeq™ CHOLESTEROL is the NGS-bases kit for the molecular profiling of Familial Hypercholesterolemia (FH).  
 AviSeq™ CHOLESTEROL allow complete characterization of 9 genes involved in Familial Hypercholesterolemia and the identification of polygenic FH SNPs and SNPs for the prediction of statin response. Familial hypercholesterolemia is a genetic disorder that affects about 1 in 250 people and increases the likelihood of having coronary heart disease at a younger age.  
 The importance of an NGS test is reflected in the timeliness of diagnosis and in the possibility of predicting the therapeutic path.



## SOLUTION

AviSeq™ CHOLESTEROL is a kit for the analysis of the FH gene through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels) of DNA extracted from body tissues (blood or others).  
 AviSeq™ CHOLESTEROL kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina.



## WORKFLOW

The AviSeq™ CHOLESTEROL kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing.  
 Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.

Table 1. List of target regions in AviSeq™ CHOLESTEROL

APOB	LDLR	PCSK9	LDLRAP1	STAP1
APOE	ABCG5	ABCG8	LIPA	

Sample per Run	
Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	16
MiSeq Nano Kit v2 (500-cycles)	16
MiSeq Micro Kit v2 (300-cycles)	68
MiSeq Kit v2 (300-cycles)	260
MiSeq Kit v2 (500-cycles)	260
MiniSeq Mid Output Kit (300-cycles)	138
iSeq 100 i1 kit (300-cycles)	68
Ion 314™ Chip	6
Ion 316™ Chip	46
Ion 318™ Chip/Ion 520™ Chip	>96

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup

Ordering Information	
Test Per Kit	Kit Reference No.
AviSeq™ CHOLESTEROL (16 test)	AVG700016
AviSeq™ CHOLESTEROL (96 test)	AVG700096
<i>For Illumina instrument</i>	
Aviseq™ Index Set 16 (16 test)	AVG404
Aviseq™ Index Set 96 (96 test)	AVG411
Aviseq™ Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
AviSeq™ Barcode Set 1-16 (96 test)	AVG502
AviSeq™ Barcode Set 17-32 (96 test)	AVG503



# AviSeq™ CLINSOME

For the analysis of the clinical exome



## PANEL

AviSeq™ CLINSOME is Allowed for the identification of molecular causes in subjects suffering from conditions not attributable to known pathologies, in subjects affected by genetically heterogeneous diseases and complex syndromic pictures.

AviSeq™ CLINSOME is a kit for the analysis of 26000 references, comprising OMIM genes, CCDS related at least 1 time to diseases, alternative transcripts, through a molecular protocol based on Next Generation Sequencing (NGS) technologies.

The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues samples.

AviSeq™ CLINSOME kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina.



## SOLUTION

Comprehensive, accurate analysis of clinically significant genes within human whole exome regions. Analyse a wide range of genetic diseases, including inherited disease related genes



## TECHNOLOGY

The AviSeq™ CLINSOME kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straightforward, capture based protocol that can be completed in as little as 36 hours, with < 3 hours hands on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.

## Sample per Run

Instrument	Sample per Run (300x)	Sample per Run (100x)
NextSeq 550 Mid-Output Kit	4	16
NextSeq 550 High-Output Kit	16	48
NovaSeq 6000 SP	32	96
NovaSeq 6000 S1	64	192

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AVISEQ™ CLINSOME (16 test)	AVG302016
AVISEQ™ CLINSOME (96 test)	AVG302096

### For Illumina instrument

AviSeq™ UDI Primers Set A (96 test)	AVG406
AviSeq™ UDI Primers Set B (96 test)	AVG407
AviSeq™ UDI Primers Set C (96 test)	AVG408
AviSeq™ UDI Primers Set D (96 test)	AVG409
AviSeq™ UDI Primers Set 16 (16 test)	AVG410

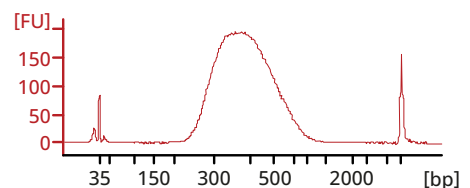


Figure 1. AviSeq™ CLINSOME library Bioanalyzer profile.



# AviSeq™ COLUNG

For the screening of Metastatic Colorectal Cancer and Non-small-cell Lung Carcinoma



## PANEL

AviSeq™ COLUNG is a novel NGS Medical Device for the molecular profiling of the Hot Spots relevant for the therapy selection in Metastatic Colorectal Cancer and non small-cell lung carcinoma.



## SOLUTION

AviSeq™ COLUNG is a kit for the analysis of the KRAS, NRAS, BRAF, EGFR and PIK3CA genes through a molecular protocol based on NGS technologies. The kit is validated for somatic analysis (SNPs, indels) of DNA extracted from cancer tissues (fresh, frozen or FFPE) or other body tissues. AviSeq™ COLUNG kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## WORKFLOW

The AviSeq™ COLUNG kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyser.

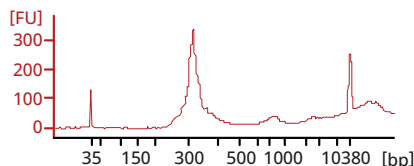


Figure 1. AviSeq™ COLUNG profiles examples

Table 1. List of target regions in AviSeq™ COLUNG

Target genes	Exons	Target genes	Exons	Target genes	Exons
KRAS	2, 3, 4	BRAF	11, 15	PIK3CA	10, 21
NRAS	2, 3, 4	EGFR	18, 19, 20, 21		

## Sample per Run

Instrument	Sample Per Run
MiSeq Nano Kit v2 (300-cycles)	7
MiSeq Nano Kit v2 (500-cycles)	7
MiSeq Micro Kit v2 (300-cycles)	30
MiniSeq Mid Output Kit (300-cycles)	60
iSeq 100 i1 kit (300-cycles)	30
Ion 314™ Chip	8
Ion 316™ Chip	36
Ion 318™ Chip/Ion 520™	80

\*The maximum number of samples per cartridge/chip estimated assuming an average depth of 5000x for somatic samples. The optimal number of samples must be empirically determined on local setups.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ COLUNG (16 test)	AVG102016
AviSeq™ COLUNG (48 test)	AVG102048
<i>For Illumina instrument</i>	
AviSeq™ Index Set 16 (16 test)	AVG404
AviSeq™ Index Set 96 (96 test)	AVG411
<i>For Ion Torrent instrument</i>	
AviSeq™ Barcode Set 1-16 (96 test)	AVG502
AviSeq™ Barcode Set 17-32 (96 test)	AVG503



# AviSeq™ COV19

For the characterization of SARS CoV 2 virus



## PANEL

- Detection of all known variants
- Optimized to identify the Omicron variant
- Sequence of the entire SARS CoV 2 genome
- Dedicated analysis software
- Hands on time < 1h



## TECHNOLOGY

AviSeq™ COV19 is a CE IVD kit for the analysis of the SARS CoV 2 genome through a molecular protocol based on NGS technologies.

The kit contains all the reagents required for the preparation of specific bi directional amplicon libraries for Illumina and Ion Torrent platforms.

This technology provides an easy to use and fast solution for characterization of SARS CoV 2 through a quick and easy workflow.



## WORKFLOW

Library preparation follows a straightforward, PCR based protocol that can be completed in as little as 5 hours, with 1 hours hands on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing.



## VALIDATION

AviSeq™ COV19 is part of an integrated solution with a dedicated proprietary pipeline for data analysis. Avicenna™ support the implementation and customization of the analysis based on the needs of the laboratory.

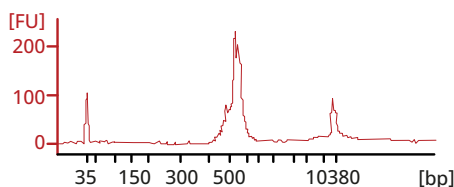


Figure 1. AviSeq™ COV19 library Bioanalyzer profile.

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (500-cycles)	3
MiSeq Kit v2 (500-cycles)	48
MiSeq Kit v3 (600-cycles)	72
Ion 314TM Chip	2
Ion 316TM Chip	16
Ion 318TM Chip	48
Ion 350TM Chip	>96
Ion PITM Chip/Ion 540TM Chip	>96

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ COV19 (16 test)	AVG212016
AviSeq™ COV19 (96 test)	AVG212096
Aviseq™ COV19 (384 test)	AVG212384
<i>For Illumina instrument</i>	
Aviseq™ Index Set 16 (16 test)	AVG404
Aviseq™ Index Set 96 (96 test)	AVG411
Aviseq™ Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
Aviseq™ Barcode Set 1-16 (96 test)	AVG502
Aviseq™ Barcode Set 17-32 (96 test)	AVG503



# AviSeq™ HEMACAN

For the analysis of complex genomic variants associated with Lymphoid and Myeloid Diseases



## PANEL

The AviSeq™ HEMACAN kit is dedicated to the analysis of complex genomic variants associated with Lymphoid and Myeloid Diseases.

AviSeq™ HEMACAN is a kit for the analysis of 137 relevant genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues (fresh, frozen, FFPE, FNA) samples.

AviSeq™ HEMACAN kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina.

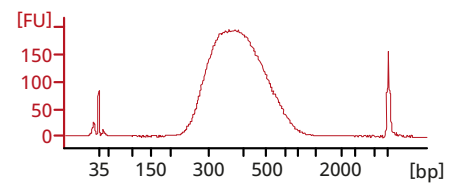


Figure 1. AviSeq™ HEMACAN library Bioanalyzer profile.



## TECHNOLOGY

The AviSeq™ HEMACAN kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.

Sample per Run	
Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	1
MiSeq Nano Kit v2 (500-cycles)	1
MiSeq Micro Kit v2 (300-cycles)	6
MiSeq Kit v2 (300-cycles)	24
MiSeq Kit v2 (500-cycles)	24
MiSeq Kit v3 (600-cycles)	48
MiniSeq Mid Output Kit (300-cycles)	12
MiniSeq High Output Kit (300-cycles)	40
iSeq 100 i1 kit (300-cycles)	6

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

Table 1. List of target regions in AviSeq™ HEMACAN

ABL1	CHEK2	GATA1	LUC7L2	RAD21	SRSF2	BIRC	IRF4	XPO1
ANKRD26	CREBBP	GATA2	MECOM	RAF1	STAG1	BTk	MAL	
ASXL1	CSF3R	GNAS	MET	RB1	STAG2	CARD11	MEF2B	
ASXL2	CSMD1	GNB1	MPL	RBBP6	STAT3	CCND1	MYD88	
ATM	CSNK1A1	HNRNPk	MYC	RPS19	STAT5B	CCND3	NFKBIE	
ATRX	CTCF	HRA5	NF1	RTEL1	TERC	CD58	NRAS	
BCOR	CUX1	IDH1	NOTCH1	RUNX1	TET	CD79A	PIM1	
BCORL1	DDX41	IDH2	NOTCH2	SAMD9	TET2	CD79B	PLCG2	
BRAF	DHX15	IKZF1	NPM1	SAMD9L	TP53	CDKN2B	POT1	
BRCE3	DNMT3A	JAK1	NRAS	SBDS	UZAF1	CHD2	PRDM1	
CALR	ELANE	JAK2	PAX5	SETBP1	WT1	CIITA	PTEN	
CBL	ETN1K1	JAK3	PDGFRA	SF3B1	ZBTB7A	CXCR4	REL	
CBLB	ETV6	KDM6A	PHF6	SH2B3	ZRSR2	EP300	SOC3	
CBLc	EZH2	KIT	PIGA	SMC1A	ARID1A	FBXW7	STAT6	
CCND2	FANCA	KMT2A	PML	SMC3	B2M	FOXO1	TCF3	
CDKN2A	FANCL	KMT2D	PPM1D	SOS1	BCL2	GNA13	TNFAIP3	
CEBPA	FLT3	KRAS	PTPN11	SRP72	BCL6	ID3	TNFRSF14	

Ordering Information	
Test Per Kit	Kit Reference No.
AviSeq™ HEMACAN (16 test)	AVG600016
AviSeq™ HEMACAN (96 test)	AVG600096
<i>For Illumina instruments</i>	
AviSeq™ UDI Primers Set A (96 test)	AVG406
AviSeq™ UDI Primers Set B (96 test)	AVG407
AviSeq™ UDI Primers Set C (96 test)	AVG408
AviSeq™ UDI Primers Set D (96 test)	AVG409
AviSeq™ UDI Primers Set 16 (16 test)	AVG410



# AviSeq™ HRD

Variants profiling and HRD score in Breast, Ovary and Prostate Cancer



## PANEL

AviSeq™ HRD is the kit for the identification of mutations in genes related to the Homologous recombination and repair (HRR) pathway.

HRR deficiency (HRD) is involved in the tumorigenesis and progression of cancer: several studies demonstrated that HRD score is a biomarker of sensitivity to platinum chemotherapy drugs.

AviSeq™ HRD is a kit for the analysis of 24 genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline and somatic analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues (fresh, frozen, FFPE, FNA) samples. AviSeq™ HRD kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina..

Table 1. List of target regions in AviSeq™ HRD

ATM	RAD51D	CHEK2	MLH1
BRCA1	RAD50	MRE11A	MSH2
BRCA2	BRIP1	NBN	MSH6
RAD54L	BARD1	PALB2	TP53
RAD50	CDK12	RAD51B	FANCL
RAD51C	CHEK1	PTEN	



## TECHNOLOGY

The AviSeq™ HRD kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.



## REFERENCES

- Kim SJ et al. Determining homologous recombination deficiency scores with whole exome sequencing and their association with responses to neoadjuvant chemotherapy in breast cancer. *Transl Oncol.* 2021
- Takaya H. et al. Homologous recombination deficiency status-based classification of high-grade serous ovarian carcinoma. *Sci Rep* 2020.

## Sample per Run

Instrument	Sample per Run	
	Germline	Somatic
MiSeq Nano Kit v2 (300-cycles)	8	0
Nano Kit v2 (500-cycles)	8	0
Micro Kit v2 (300-cycles)	32	2
MiniSeq Mid Output Kit (300-cycles)	64	4
iSeq 100 i1 kit (300-cycles)	32	2

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ HRD (16 test)	AVG106016
AviSeq™ HRD (96 test)	AVG106096
<i>For Illumina instruments</i>	
AviSeq™ UDI Primers Set A (96 test)	AVG406
AviSeq™ UDI Primers Set B (96 test)	AVG407
AviSeq™ UDI Primers Set C (96 test)	AVG408
AviSeq™ UDI Primers Set D (96 test)	AVG409
AviSeq™ UDI Primers Set 16 (16 test)	AVG410



# AviSeq™ MICROBIOME

For the analysis of the hypervariable regions of the bacterial 16S rDNA gene



## PANEL

AviSeq™ MICROBIOME is a novel NGS Medical Device for the analysis of the hyper variable regions of the bacterial 16 S rDNA gene.



## SOLUTION

AviSeq™ MICROBIOME is a kit for the analysis and phylogenetic characterization of several microbial communities through a molecular protocol based on NGS technologies (Table 1).

The kit is validated for analysis of DNA extracted from fecal, pulmonary, oral, vaginal samples and from surface and soil samples.

AviSeq™ MICROBIOME kit contains all reagents required for the preparation of a specific bidirectional amplicons library designed for NGS analysis using Illumina, Ion Torrent or Nanopore sequencers



## WORKFLOW

The AviSeq™ MICROBIOME kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded into a flow cell for sequencing.

**SPECIFICITY** > 99%

**SENSITIVITY** > 99%

**COVERAGE (>30X)** > 99%

Table 1. List of target regions in AviSeq™ MICROBIOME

HUMAN microbiome	V1-V2	Vaginal microbiome
	V3-V4	Intestinal Microbiome
	V5-V6	Lactose enzymes
SURFACES	V7 - V9	Surface and soil microorganisms

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (500-cycles)	7
MiSeq Kit v2 (500-cycles)	27
MiSeq Kit v3 (600-cycles)	45
NextSeq 550 Mid-Output Kit Ion	234
510TM Chip	5
Ion 318TM Chip/Ion 520TM Chip	12
Ion 530TM Chip	37
Ion PiTM Chip/Ion 540TM Chip	250

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ MICROBIOME (16 test)	AVG800016
AviSeq™ MICROBIOME (48 test)	AVG800048
<i>For Illumina instrument</i>	
AviSeq™ Index Set 16 (16 test)	AVG404
AviSeq™ Index Set 96 (96 test)	AVG411
AviSeq™ Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
AviSeq™ Barcode Set 1-16 (96 test)	AVG502
AviSeq™ Barcode Set 17-32 (96 test)	AVG503



# AviSeq™ ONCO 50

Hereditary and somatic variants profiling in Breast and Ovary Cancer



## PANEL

AviSeq™ ONCO 50 is kit for the identification of mutations in genes related to Breast and Ovary cancer, Familial adenomatous polyposis (FAP) and hereditary nonpolyposis colorectal cancer (HNPCC), two syndromes of colorectal cancer predisposition, inherited in an autosomal dominant fashion.

AviSeq™ ONCO 50 allows the search for variants linked to a defect in homologous recombination repair (HRD), similarly to pathogenic variants in BRCA1 or BRCA2. The defect in HR causes error-prone DNA repair pathways in the cell, resulting in increased genomic instability, which may be responsible for susceptibility to poly-(ADP)-ribose polymerase (PARPis) inhibitors.

AviSeq™ ONCO 50 is a kit for the analysis of 50 genes through a molecular protocol based on Next Generation Sequencing (NGS) technologies. The kit is validated for germline and somatic analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues (fresh, frozen, FFPE, FNA) samples.

AviSeq™ ONCO 50 kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina sequencers.



## TECHNOLOGY

The AviSeq™ ONCO 50 kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.



## WORKFLOW

Library preparation follows a straightforward, capture based protocol that can be completed in as little as 36 hours, with < 3 hours hands on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible illumina sequencers.



## VALIDATION

Todemonstrate assay capabilities, clinical samples were run in a clinical setting DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer Profile of the prepared libraries in Figure.

Table 1. List of target regions in AviSeq™ ONCO 50

ATM	CHEK2	MUTYH	RAD51D	PIK3CA	SMARCB1	ALK	CDKN1C
APC	EPCAM	NBN	STK11	PMS2CL	NF2	EGFR	
BARD1	MLH1	PALB2	TP53	XRCC2	PTCH1	RB1	
BRCA1	MSH2	PMS2	WRN	NF1	LZTR1	CDKN2A	
BRCA2	XPC	PTEN	HRAS	SPRED1	CTNNB1	CDK4	
BRIP1	SMAD4	RAD50	FAM175A	MEN1	KRAS	FLCN	
CDH1	MSH6	RAD51C	MRE11	RET	ERBB2	RUNX1	

## Sample per Run

Instrument	Sample per Run	
	Germline	Somatic
MiSeq Nano Kit v2 (300-cycles)	4	0
• Nano Kit v2 (500-cycles)	4	0
• Micro Kit v2 (300-cycles)	16	1
• Kit v2 (300-cycles)	56	3
• Kit v2 (500-cycles)	56	3
• Kit v3 (600-cycles)	96	6
MiniSeq Mid Output Kit (300-cycles)	32	1
• High Output Kit (300-cycles)	96	6
iSeq 100 i1 kit (300-cycles)	16	1

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ ONCO 50 (16 test)	AVG105016
AviSeq™ ONCO 50 (96 test)	AVG105096
<i>For Illumina instruments</i>	
AviSeq UDI Primers Set A (96 test)	AVG406
AviSeq UDI Primers Set B (96 test)	AVG407
AviSeq UDI Primers Set C (96 test)	AVG408
AviSeq UDI Primers Set D (96 test)	AVG409
AviSeq UDI Primers Set 16 (16 test)	AVG410





# AviSeq™ PANCAN SD

For the screening of solid tumors.



## PANEL

AviSeq™ PANCAN SD is a novel NGS medical device for the molecular profiling of the variants involved in onset and progression of solid tumors.



## SOLUTION

AviSeq™ PANCAN SD is a kit for the analysis of 96 genes related to several solid tumors, through a molecular protocol based on NGS technologies. The kit is validated for germline analysis (SNPs, indels, CNVs) of DNA extracted from blood or body tissues samples.

AviSeq™ PANCAN SD kit contains all reagents required for the preparation of the capture of specifically designed probes and for the NGS analysis using Illumina.



## WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 24 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencers.



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

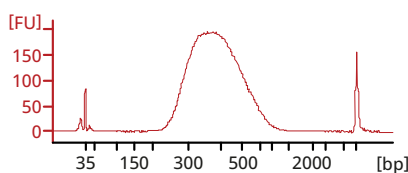


Figure 1. AviSeq™ PANCAN SD library Bioanalyzer profile.

## Sample per Run

Instrument	Sample per Run
MiSeq Kit v2 (300-cycles)	16
MiSeq Kit v2 (500-cycles)	16
MiSeq Kit v3 (600-cycles)	32
MiniSeq Mid Output Kit (300-cycles)	8
MiniSeq High Output Kit (300-cycles)	32

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ PANCAN SD (16 test)	AVG107016
AviSeq™ PANCAN SD (96 test)	AVG107096
<i>For Illumina instruments</i>	
UDI Primers Set A (96 test)	AVG406
UDI Primers Set B (96 test)	AVG407
UDI Primers Set C (96 test)	AVG408
UDI Primers Set D (96 test)	AVG409
UDI Primers Set 16 (16 test)	AVG410

Table 1. List of target regions in AviSeq™ PANCAN SD

ABL1	CDKN2A	FGFR3	NRAS	TP53	P13K	CDH1	MLP
ALK	CHEK2	FLT3	PALB2	VHL	EGFR	CSF1R	NPM1
APC	CHEK1	HRAS	PIK3CA	BRCA1	PDL1 (CD274)	DDR2	PDGFRA
ARID1A	CSF3R	IDH1	PTEN	BRCA2	KRAS	ERBB2	PTPN11
ATM	CTNNB1	IDH2	RAD51B	HER2	NOTCH1	ERBB4	STK11
ATR	DNMT3A	JAK2	RAD51C	MET	NOTCH2	EZH2	SMAD4
ATRX	FANCA	JAK3	RAD51D	NTRK1	NOTCH3	FOXL2	SMARCB1
BARD1	FANCL	KIT	RAD54L	NTRK2	NOTCH4	GNA11	SMO
BRAF	FBXW7	KMT2A	RB1	NTRK3	AKT1	GNAQ	SRC
BRIP1	EML4	MAP2K1	RET	HRAS	PIK3CA	GNAS	TSC1
CBL	FGFR1	MLH1	ROS1	KRAS	TSHR	HNFB1A	VHL
CDK12	FGFR2	MSH6	TET2	NRAS	TERT	KDR	



# AviSeq™ RESPASENSE

For the screening of susceptibility to respiratory infectious diseases



## PANEL

AviSeq™ RESPASENSE (Respiratory Infectious Diseases Susceptibility) is a kit designed for large population screening through a molecular protocol based on NGS technologies. The resulting genetic profile evaluates both genetic variants in the infection pathway (predisposition or protection against infection), and variants of the genes involved in the immune response to infection (predisposition or protection against a worse outcome). The kit is validated for analysis of DNA extracted from different body tissues (blood, saliva, etc.). AviSeq™ RESPASENSE kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## SOLUTION

AviSeq™ RESPASENSE (Respiratory Infectious Diseases Susceptibility) is a kit designed for large population screening through a molecular protocol based on NGS technologies. The resulting genetic profile evaluates both genetic variants in the infection pathway (predisposition or protection against infection), and variants of the genes involved in the immune response to infection (predisposition or protection against a worse outcome). The kit is validated for analysis of DNA extracted from different body tissues (blood, saliva, etc.). AviSeq™ RESPASENSE kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## WORKFLOW

The AviSeq™ RESPASENSE kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis.

Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded for sequencing.

Table 1. List of target regions in AviSeq™ RESPASENSE

		GENE	RSID
susceptibility/ resistance	aspecific	ABO (depends on group)	rs657152
		DDP4 (associated with resistance)	rs13015258 rs117888248 rs116302758 rs56179129 rs115450134
		CD147 (BSG)	rs201850688 rs11551906 rs144824657 rs41276870
	-specific for COVID19	CCR5 (associated with resistance)	rs333
		ACE1	rs4341
		ACE2 (associated with resistance to infection)	K31R, N33I, H34R, E35K, E37K, D38V, Y50F, N51S, M62V, K68E, F72V, Y83H, G326E, G352V, D355N, Q388L, and D509Y
GWAS	TMPRSS2 (associated with increased susceptibility)	rs2070788 rs383510 rs200291871 rs75603675 rs61735791 rs114363287 rs12329760	
	GLL5, GNAZ, RSPH14, RAB36 and BCR	rs73166864	
outcome (associated with worst prognosis)	IVNS1ABP, SWT1	rs6668622	
	ApoE	rs429358-C-C (e4e4)	
	IFITM3	rs12252 rs6598045	
	SLC6A20, LZTFL1, CCR9, FYCO1, CXCR 6, XCR1	rs11385942 rs73064425	
	OAS3	rs10735079	
	TMEM189 UBE2V1	rs6020298A	
	DPP9	rs2109069	
	PCSK3 (associate with )	rs16944971 rs780909157 rs201551785 rs769208985 rs1236237792	
SRRM1, IVNS1ABP (hospitalization)	rs111972040		

Figure 1: workflow





# AviSeq™ RESPASENSE

For the screening of susceptibility to respiratory infectious diseases



## DATA ANALYSIS

First step of data analysis is performed with our proprietary platform.



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer

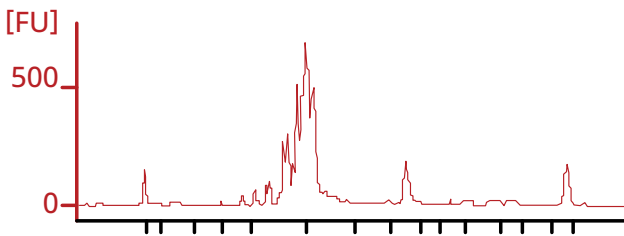


Figure 2. AviSeq™ RESPASENSE library Bioanalyzer profile.

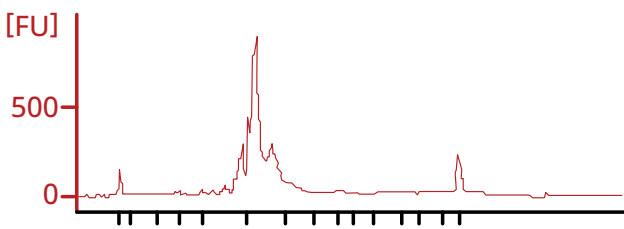


Figure 3. AviSeq™ RESPASENSE library Bioanalyzer profile.



## REFERENCES

1. Anastassopoulou C et al, Human Genomics 2020;
2. Pairo Castineira et al, Nature 2020;
3. Ellinghaus et al, Nature 2020;
4. Zhang et al, Science 2020

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	2
MiSeq Nano Kit v2 (500-cycles)	2
MiSeq Micro Kit v2 (300-cycles)	8
MiSeq Kit v2 (300-cycles)	32
MiSeq Kit v2 (500-cycles)	32
MiSeq Kit v3 (600-cycles)	48
MiniSeq Mid Output Kit (300-cycles)	16
MiniSeq High Output Kit (300-cycles)	48
iSeq 100 i1 kit (300-cycles)	8
NextSeq 550 Mid-Output Kit	288
Ion 314™ Chip	8
Ion 316™ Chip	36
Ion 318™ Chip/Ion 520™ Chip Ion	80
530™ Chip	>96
Ion Pi™ Chip/Ion 540™ Chip	>96

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ RESPASENSE (16 test)	AVG304016
AviSeq™ RESPASENSE (96 test)	AVG304096

### For Illumina instrument

Aviseq™ Index Set 16 (16 test)	AVG404
Aviseq™ Index Set 96 (96 test)	AVG411
Aviseq™ Index Set 384 (384 test)	AVG405

### For Ion Torrent instrument

Aviseq™ Barcode set 1-16 (96 test)	AVG502
Aviseq™ Barcode Set 17-32(96 test)	AVG503



# AviSeq™ RESPASENSE PLUS

For the screening of susceptibility to respiratory infectious diseases



## PANEL

Clinicians around the world have found that different people of the same gender and age had different outcomes when infected with SARS COV 2 even within the same family. Information on the genetic profile of the host (the infected subject) is becoming increasingly important, with the identification of specific genetic markers that can support the preventive identification of subjects at high or low risk.

According to recent studies, there is a strict correlation between genetic variants and:

- susceptibility to viral infection
- propensity to develop harmful pulmonary inflammation
- persistence of positivity
- vaccine resistance or development of adverse events

There are currently no solutions on the market that allow genetic profile screening. Avicenna™ has developed a genetic test to meet this need: AviSeq RESPASENSE PLUS (for Respiratory Infectious Diseases Susceptibility). The tool is a complete solution from DNA extracted from blood or buccal swab, up to creation of a report.



## SOLUTION

Avicenna™ has a consolidated experience in the development of CE IVD genetic tests for the study of genetic predisposition to genetic diseases. Thanks to the collaboration with two university hospitals, Tor Vergata and Bambin Gesù, Avicenna™ is correlating anonymized key clinical data with the genetic markers obtained, to elaborate a genetic risk score.

AviSeq™ RESPASENSE PLUS does not stop at SARS COV 2 but the genetic markers contained in the kit correspond to variants of the immune system target also of other RNA viruses, such as the flu virus. This makes the project larger than the current pandemic and AviSeq™ RESPASENSE PLUS product a powerful tool applicable in the future to the prevention of infectious respiratory diseases, including the pandemics of the 2000s and new pandemics.

AviSeq™ RESPASENSE PLUS is a kit designed to screen genes involved in the infection pathway (predisposition or protection against infection), and in the host immune response to infection (predisposition or protection against a worse outcome).

The kit is validated for analysis of DNA extracted from different body tissues (blood, saliva, buccal swab, etc.).

AviSeq™ RESPASENSE PLUS kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina.



## WORKFLOW

AviSeq™ RESPASENSE PLUS kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, capture-based protocol that can be completed in 1 day and a half, with a total hands-on time of 5 hours. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina sequencer.

Table 1. List of target regions in AviSeq™ RESPASENSE PLUS

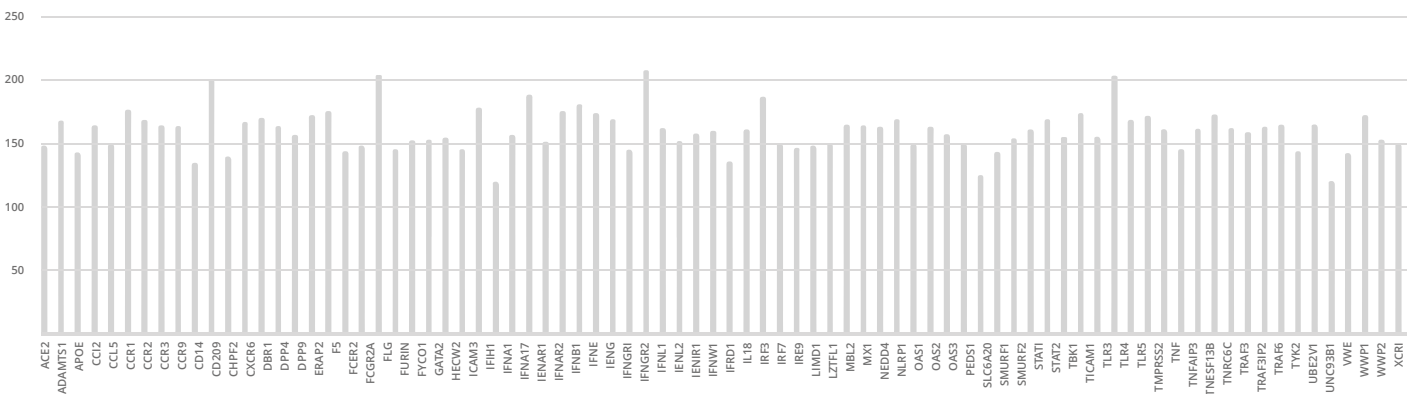
CDKN2A	FGFR3	NRAS	TP53	
ALK	CHEK2	FLT3	PALB2	VHL
APC	CHEK1	HRAS	PIK3CA	BRCA1
ARID1A	CSF3R	IDH1	PTEN	BRCA2
ATM	CTNNB1	IDH2	RAD51B	HER2
ATR	DNMT3A	JAK2	RAD51C	MET
ATRX	FANCA	JAK3	RAD51D	NTRK1
BARD1	FANCL	KIT	RAD54L	NTRK2
BRAF	FBXW7	KMT2A	RB1	NTRK3
BRIP1	EML4	MAP2K1	RET	HRAS
CBL	FGFR1	MLH1	ROS1	KRAS
CDK12	FGFR2	MSH6	TET2	NRAS



# AviSeq™ RESPASENSE PLUS

For the screening of susceptibility to respiratory infectious diseases

Figure1: Mean coverage on target at 50x >99%



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

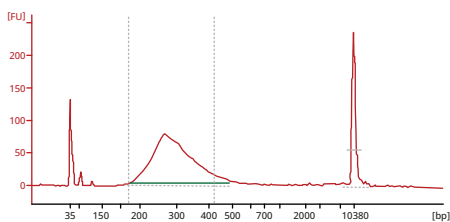


Figure 1. AviSeq™ RESPASENSE PLUS library Bioanalyzer profile.

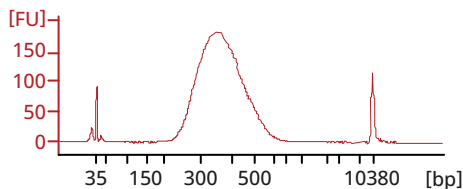


Figure 1. AviSeq™ RESPASENSE PLUS library Bioanalyzer profile.

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	2
MiSeq Nano Kit v2 (500-cycles)	2
MiSeq Micro Kit v2 (300-cycles)	8
MiSeq Kit v2 (300-cycles)	32
MiSeq Kit v2 (500-cycles)	32
MiSeq Kit v3 (600-cycles)	48
MiniSeq Mid Output Kit (300-cycles)	16
MiniSeq High Output Kit (300-cycles)	48
iSeq 100 i1 kit (300-cycles)	8

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ RESPASENSE PLUS (16 test)	AVG301016
AviSeq™ RESPASENSE PLUS (96 test)	AVG301096
<i>For Illumina instruments</i>	
UDI Primers Set A (96 test)	AVG406
UDI Primers Set B (96 test)	AVG407
UDI Primers Set C (96 test)	AVG408
UDI Primers Set D (96 test)	AVG409
UDI Primers Set 16 (16 test)	AVG410



## REFERENCES

- Anastassopoulou C et al, Human Genomics 2020;
- Païro-Castineira et al, Nature 2020;
- Ellinghaus et al, Nature 2020;
- Zhang et al, Science 2020



# AviSeq™ THALASEMIA

For the screening variants causing alpha and beta Thalassemia

## PANEL

AviSeq™ THALASEMIA is a novel NGS Medical Device for the molecular profiling of variants causing alpha and beta THALASEMIA. THALASEMIA is the name for a group of inherited characterized by abnormal hemoglobin production.

## SOLUTION

AviSeq™ THALASEMIA is a kit for the analysis of the HBB, HBA1, HBA2, HBD, HBG1, HBG2, HS-40 and LCRB genes through a molecular protocol based on NGS technologies.

The kit is validated for germline analysis (SNPs, indels) of DNA extracted from blood samples.

AviSeq™ THALASEMIA kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.

## WORKFLOW

The AviSeq™ THALASEMIA kit is part of a DNA-to-variant solution that offers streamlined content, easy-to perform library preparation, push-button sequencing systems, and simplified data analysis.

Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing.

Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.

## TARGET REGIONS

Entire genes:

- HBB, HBA1, HBA2
- Alpha globin cluster related
  - POLR3K, HS-40, HBZ, HBZP1, HBAP1, HBA2,
  - HBA1, HBQ1, LUC7L, AXIN1
- Beta globin cluster related
  - LCRB, HBG2, HBG1, HBD, HBB, 3'HS1

Table 1. List of target regions in AviSeq™ THALASEMIA

Alpha globin deletions	Beta globin deletions
--SEA	Chinese
--FIL	Filipino
--THAI	Yunnanese
-(a)20.5	Taiwanese
--MED	SEA-HPFH
-(a)21.9	Hb-Lepore Boston
-(a)27.6	Hb-Lepore Baltimore
-(a)3.7	290bp-del
-(a)4.2	B619-del
	SiSicilia

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	6
MiSeq Nano Kit v2 (500-cycles)	6
MiSeq Micro Kit v2 (300-cycles)	28
MiSeq Kit v2 (300-cycles)	>96
MiSeq Kit v2 (500-cycles)	>96
MiSeq Kit v3 (600-cycles)	>96
MiniSeq Mid Output Kit (300-cycles)	57
MiniSeq High Output Kit (300-cycles)	>96
iSeq 100 i1 kit (300-cycles)	28
Ion 314TM Chip	4
Ion 316TM Chip	18
Ion 318TM Chip/Ion 520TM Chip	35
Ion 530TM Chip	>96

\*The estimated maximum number of samples per cartridge / chip The optimal number of samples can be empirically estimated on the local setup

## Ordering Information

Test Per Kit	Kit Reference No.
AVISEQ™ THALASEMIA (16 test)	AVG601016
AVISEQ™ THALASEMIA (96 test)	AVG601096
<i>For Illumina instruments</i>	
AviSeq Index Set 16 (16 test)	AVG404
AviSeq Index Set 96 (96 test)	AVG411
AviSeq Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instruments</i>	
Aviseq™ Barcode Set 1-16 (96 test)	AVG502
Aviseq™ Barcode Set 17-32 (96 test)	AVG503



# AviSeq™ THYRO

For the screening of Thyroid cancer



## PANEL

AviSeq™ THYRO is a novel NGS Medical Device for the molecular profiling of the variants involved in the development of Thyroid cancer.



## SOLUTION

AviSeq™ THYRO is a kit for the analysis of KRAS, NRAS, BRAF, EGFR, CTNNB, AKT1, PTEN, CDKN2A, HRAS, TSHR, NOTCH, TP53, PIK3CA genes and hTERT promoter through a molecular protocol based on NGS technologies. The kit is validated for somatic analysis (SNPs, indels) of DNA extracted from tissues (fresh, frozen, FFPE, FNA, etc.). AviSeq™ THYRO kit contains all reagents required for the preparation of a specific bidirectional library of amplicons designed for the NGS analysis using Illumina or Ion Torrent sequencers.



## WORKFLOW

The AviSeq™ THYRO kit is part of a DNA-to-variant solution that offers streamlined content, easy-to-perform library preparation, push-button sequencing systems, and simplified data analysis. Library preparation follows a straightforward, PCR-based protocol that can be completed in as little as 5 hours, with < 1.5 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Illumina or Ion Torrent sequencers.



## VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.

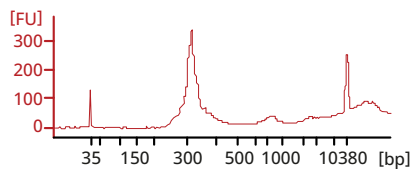


Figure 1. AviSeq™ THYRO library Bioanalyzer profile.

Table 1. List of target regions in AviSeq™ THYRO

Target Genes	Exons	Target Genes	Exons	Target Genes	Exons	Target Genes	Exons	Target Genes	Exons
KRAS	2, 3, 4	EGFR	18, 19, 20, 21	PIK3CA	10, 21	HRAS	2,3	TP53	4, 5, 6, 7, 8, 9
NRAS	2, 3, 4	CTNNB1	1	CDKN2A	1, 2	TSHR	6, 8, 9	hTERT	promoter
BRAF	15	AKT1	1	PTEN	5,6,7,8	NOTCH	26, 27		

## Sample per Run

Instrument	Sample per Run
MiSeq Nano Kit v2 (300-cycles)	5
MiSeq Nano Kit v2 (500-cycles)	5
MiSeq Micro Kit v2 (300-cycles)	21
MiSeq Kit v2 (300-cycles)	80
MiSeq Kit v2 (500-cycles)	80
MiSeq Kit v3 (600-cycles)	>96
MiniSeq Mid Output Kit (300-cycles)	42
MiniSeq High Output Kit (300-cycles)	>96
iSeq 100 i1 kit (300-cycles)	21
Ion 314™ Chip	2
Ion 316™ Chip	11
Ion 318™ Chip/Ion 520™ Chip Ion 530™ Chip	22
Ion PITM Chip/Ion 540™ Chip	>96

\*The estimated maximum number of samples per cartridge / chip. The optimal number of samples can be empirically estimated on the local setup.

## Ordering Information

Test Per Kit	Kit Reference No.
AviSeq™ THYRO (16 test)	AVG103016
AviSeq™ THYRO (48 test)	AVG103048
<i>For Illumina instrument</i>	
Index Set 16 (16 test)	AVG404
Index Set 96 (96 test)	AVG411
Index Set 384 (384 test)	AVG405
<i>For Ion Torrent instrument</i>	
BARCODE Set 1 -16 (96 test)	AVG502
BARCODE Set 17-32 (96 test)	AVG503



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