Altium

DIAGNOSTIC KITS

Portfolio 2023

healthincode

Diagnostic kits

> Health in Code designs and manufactures disease diagnostic kits for use in specialized laboratories

ONCOLOGY _Hereditary cancer

Hereditary Plus OncoKitDx

CE IVD

NGS panel that analyzes 101 genes associated with the most frequent types of hereditary cancer: breast, ovarian, colorectal, uterine, melanoma, kidney, prostate, pancreatic, multiple endocrine neoplasia (MEN), pheochromocytoma, paraganglioma, and retinoblastoma. For *in vitro* diagnostic use.

Characteristics

- NGS panel for the study of the main genes associated with hereditary cancer.
- Detection of SNVs, indels, ALUs, and CNVs.
- Bioinformatics analysis with the Data Genomics software and semiautomated generation of reports.
- STIDs can be included: Integrated sample identification and tracking system.
- Coverage: 99% of bases with coverage >20x
- The uniformity of bases covered at 20% of the mean coverage is 98.8%.
- Sensitivity and specificity: >99%.
- Repeatability and reproducibility: >99%.
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked kit and analysis software.

IMG-399 Hereditary Plus OncoKitDX [101 genes]

ABRAXAS1 (FAM175A)	*BARD1*	*CHEK2*	*GALNT14*	*MET*	*NTHL1*	*RAD50*	*SDHC*	*TERT*
ACD	*BLM*	*CTNNA1*	*GDNF*	*MITF*	*PALB2*	*RAD51*	*SDHD*	*TMEM127*
AIP	*BMPR1A*	*CYLD*	*GEN1*	*MLH1*	*PHOX2B*	*RAD51B*	*SEC23B*	*TP53*
AKT1	*BRCA1*	*DICER1*	*GREM1*	*MLH3*	*PIK3CA*	*RAD51C*	*SLX4*	*TSC1*
ALK	*BRCA2*	*EPCAM (incl. 3' UTR)*	*HABP2*	*MRE11A*	*PMS2*	*RAD51D*	*SMAD4*	*TSC2*
APC (incl. 5' UTR)	*BRIP1*	*FANCC*	*HOXB13*	*MSH2*	*POLD1*	*RB1*	*SMARCA4*	*VHL*
ATM	*CDC73*	*FANCG*	*KIF1B*	*MSH3*	*POLE*	*RECQL4*	*SMARCB1*	*WT1*
ATR	*CDH1*	*FANCM*	*KLLN*	*MSH6*	*POT1*	*RET*	*SMARCE1*	*XRCC2*
AXIN2	*CDK4*	*FH*	*LZTR1*	*MUTYH*	*PRKAR1A*	*RINT1*	*SPRED1*	
BAP1	*CDKN1B*	*FLCN*	*MAX*	*NBN*	*PRSS1*	*SDHA*	*STK11*	
	CDKN1C	*GALNT12*	*MC1R*	*NF1*	*PTCH1*	*SDHAF2*	*SUFU*	
	CDKN2A		*MEN1*	*NF2*	*PTEN*	*SDHB*	*TERF2IP*	

Target genes

Technical specifications

- Compatible sequencing platforms: Illumina MiSeq and NextSeq 500/550Dx System.
- Number of reactions: 48.
- Number of samples per run:
  - MiSeq V2 300 cycles: 16 samples.
  - NextSeq Mid Output v2.5 kit (300 cycles): 32 samples.
- Sequencing: Paired-end (2 x 150 cycles).
- Sample type: DNA from peripheral blood and saliva.
- Amount of input DNA: 50-100 ng.
- Mean coverage: >350X.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**
- **Moreover, other intergenic regions of interest** for the calculation of CNV calls in the *EPCAM* (3' UTR) and *MSH2* (5' UTR) genes have been included, as well as **50 intronic regions to cover hotspots** described in the *APC, ATM, ATR, BRCA1, BRCA2, CHEK2, FH, LZTR1, MET, MLH1, MSH2, NF1, PMS2, PTEN, RB1, RET, SDHB, STK11,* and *TERT* genes.

Other kits for hereditary cancer

Technique	Reference	Kit name	Description	RXNs	
Next-Generation Sequencing	IMG-313	BRCA Plus OncoKitDx	Molecular genetic screening of the *BRCA1 and BRCA2 genes* for breast and ovarian cancer with amplicon-based NGS	16	CE IVD
Next-Generation Sequencing	IMG-326	Hereditary OncoKitDx (without index)	Analysis of 50 genes associated with predisposition to hereditary cancer, without index case	48	CE IVD
Next-Generation Sequencing	IMG-317	TP53 OncoKitDx	Molecular genetic analysis of the *TP53* gene with amplicon-based NGS	48	CE IVD

ONCOLOGY _Solid tumors

Health in Code has developed the following kits to screen for the main genomic biomarkers with recognized clinical value that have been associated with the most frequent solid tumors:

Offered kits:

IMG-365 Action OncoKitDx (Automatic)

CE IVD

Technique: Next-Generation Sequencing
RXNs: 24

IMG-419 Action ST OncoKitDx

RUO

Technique: Next-Generation Sequencing
RXNs: 24

Common characteristics

- It uses a single DNA sample to detect SNVs, indels, gene fusions, CNVs, pharmacogenetic variants, and microsatellite instability analysis (MSI).
- Bioinformatics analysis with the Data Genomics software and semiautomated generation of reports, including functional and clinical classification of the identified variants.
- STIDs can be included: Integrated sample identification and tracking system.
- Based on high sensitivity RNA-probe technology with UMI barcoding (unique molecular identifiers).
- Sensitivity, specificity, repeatability, and reproducibility of 99%.
- Limit of detection: SNVs, indels, and structural variants: 5%; CNVs: 3 copies for copy number gain and 1 copy for deletions.
- CNV analysis requires that a positive control (REF: IMG-368) be used at least once.
- CE-IVD-marked analysis software.
- Compatible sequencing platforms: Illumina NextSeq 500/550Dx.
- Number of reactions: 24.
- Sequencing: Paired-end (2 x 75 cycles).
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**

Panels composition

IMG-365 Action OncoKitDx	IMG-419 Action ST OncoKit
Sequencing of all exonic regions of 55 genes: *ALK, ARID1A, ATM, ATRX, BAP1, BRAF, BRCA1, BRCA2, CDH1, CTNNB1, CHEK2, EGFR, ERBB2/HER2, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, GNA11, GNAQ, H3F3A, HIST1H3B, HIST1H3H, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MET, MLH1, MSH2, MSH6, MTOR, MYC, NRAS, NTRK1, NTRK2, NTRK3, PALB2, PBRM1, PDGFRA, PIK3CA, PMS2+5'UTR, PTEN, POLD1, POLE, RET, ROS1, SDHA, SDHB, SDHD, TERT+5'UTR, TP53, VHL*	Sequencing of all exonic regions of 56 genes: *ALK, ARID1A, ATM, ATRX, BAP1, BARD1, BRAF, BRCA1, BRCA2, BRIP1, CHEK2, CDH1, CTNNB1, EGFR, ERBB2, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, GNA11, GNAQ, H3F3A, HIST1H3B, HIST1H3H, HRAS, IDH1, IDH2, KIT, KRAS, MAP2K1, MET, MLH1, MSH2, MSH6, MTOR, MYC, NRAS, PALB2, PDGFRA, PIK3CA, PMS2 + 5'UTR, PTEN, POLD1, POLE, RB1, RAD51C, RAD51D, RET, ROS1, SDHA, SDHB, SDHD, TERT + 5'UTR, TP53, VHL*
Sequencing of hotspot regions in the TSC1 and TSC2 genes (36 regions in total) and the E17K variant in AKT1.	Sequencing of hotspot regions in the TSC1 and TSC2 genes (36 regions in total), the E17K variant in AKT1, and seven regions in *NTRK1* and *NTRK3*.
Action ST OncoKit contains all exonic regions of the *BARD1, BRIP, RB1, RAD51C, and RAD51D genes. The NTRK1, NTRK2, NTRK3, and PBRM1 genes have been removed, though* seven hotspots in *NTRK1* and *NTRK3* were kept as part of the design.	
Capture of 11 fusion genes with any possible rearrangement (including previously reported intronic breakpoint regions): *ALK, ATP1B1, BRAF, EGFR, ETV6, FGFR2, FGFR3, NTRK1, NTRK2, RET, ROS1*	Capture of 10 fusion genes with any possible rearrangement. Action ST OncoKit includes intronic regions containing breakpoint sites more commonly reported in the literature. The included genes and covered regions are the following: *ALK* (introns, 17, 18, and 19), *BRAF* (introns 7, 8, 9, and 10), *EGFR* (introns 7, 23, 24, 25, and 26), *ETV6-NTRK3* (*ETV6* introns 4 and 5), *FGFR2* (intron 17 and 3' UTR), *FGFR3* (intron 17 and 3' UTR), *NTRK1* (introns 9, 10, 11, 12, and 13), *NTRK2* (introns 10, 11, 12, and 15), *RET* (introns 7, 9, 10, and 11), and *ROS1* (introns 17, 26, 28, 31, 32, 33, 34, and 35).
Action ST OncoKit maintains the design of the *EGFR, ETV6, FGFR2*, and *FGFR3 genes*. The following summary shows the genes and introduced changes:	
ALK	Added I17 and I18
ATP1B1	Gene removed
BRAF	Added I11
NTRK1	Removed I8 and added I13
NTRK2	Added I10 and I11
ROS1	Added I17, I26, and I28
Microsatellite instability (MSI) analysis	
Detection of CNVs throughout the whole genome (detection of hypo- and hyperploidy) or in an entire gene included in the panel. Validation of results and detection of copy-neutral LOH across the genome with a low-density SNP array.	
Detection of pharmacogenetics-related variants in ***DPYD, UGT1A1, CYP2D6, MTHFR, TPMT, and CYP2C9.***	

Differences between kits

	IMG-365 Action OncoKitDx	IMG-419 Action ST OncoKit
CE-IVD mark	Yes	No
Sample type	DNA from peripheral blood and fresh, frozen, and paraffin-embedded tissue	DNA from peripheral blood and paraffin-embedded tissue
Amount of input DNA	50 – 200 ng	10 – 50 ng
Average raw coverage	2500X	3500X
Average coverage after UMI analysis	1600X	2300X
Bases covered at a depth greater than 100X	>99.3	99.4
Uniformity of bases covered at more than 20% of the average coverage	0.989	0.978
Number of samples in MID output sequencing mode	8 – 12	8
Quality standards	Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials	Compliant with quality specifications by the ISO 9001 and ISO 14001 standards regarding manufacturing materials.

HRR OncoKitDx

RUO

Sequencing of genes involved in homologous recombination repair (HRR) and microsatellite instability (MSI) in solid tumors.

Characteristics

- NGS panel and software for the testing of genes involved in homologous recombination repair (HRR) and microsatellite instability (MSI) analysis in solid tumors.
- SNV and indel detection.
- Detection of CNVs in the selected genes, as well as gains and losses across the whole genome.
- UMI molecular barcoding, which increases the sensitivity of the bioinformatic analysis.
- Limit of detection: SNVs and indels: 5%; CNVs: 3 copies for copy number gain and 1 copy for deletions.
- Bioinformatic analysis with the Data Genomics software.
- STIDs can be included: Integrated sample identification and tracking system.
- The coverage (at a depth >200X) is 97.4%. The sensitivity, specificity, repeatability, and reproducibility of this test are greater than 99%. Uniformity of coverage of bases covered at >20X is 96.6%.
- Compliant with quality specifications by the ISO 9001 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked analysis software.

Technical specifications

- Compatible sequencing platforms: Illumina NextSeq500/550 and Illumina NextSeq 550Dx.
- Number of reactions: 24.
- Number of samples per run:
  - Mid Output v2.5 kit (150 cycles): 16 samples.
  - High Output v2.5 kit (150 cycles): 32 samples.
- Sequencing: Paired-end (2 x 75 cycles).
- Sample type: DNA from peripheral blood or formalin-fixed paraffin-embedded tumor tissue.
- Recommended amount of DNA: 10-100 ng.
- Analyzed genomic region: 400 Kb.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**
- CNV analysis requires that a positive control (REF: IMG-368) be used at least once.

Panel composition

IMG-400 HRR OncoKit [38 genes]

ATM	*BRCA1*	*CHEK2*	*FANCB*	*FANCF*	*FANCM*	*NBN*	*RAD51*	*RAD52*
ATR	*BRCA2*	*ERCC3*	*FANCC*	*FANCG*	*GEN1*	*PALB2*	*RAD51B*	*RAD54L*
ATRX	*BRIP1*	*FAM175A (ABRAXAS1)*	*FANCD2*	*FANCI*	*HDAC2*	*PPP2R2A*	*RAD51C*	*RPA1*
BARD1	*CDK12*	*FANCE*	*FANCL*	*MRE11A*	*RAD50*	*RAD51D*	*WRN*	
BLM	*CHEK1*	*FANCA*						

Target genes: SNVs and INDELs

Other kits for solid tumors

Technique	Reference	Kit name	Description	RXNs	
Next-Generation Sequencing	IMG-313	BRCA Plus OncoKitDx	Molecular genetic screening of the *BRCA1* and *BRCA2* genes for breast and ovarian cancer with amplicon-based NGS	16	CE IVD
Fragment analysis (PCR)	IMG-314	MSI OncoKitDx	Detection of microsatellite instability	48	CE IVD
Next-Generation Sequencing	IMG-317	TP53 OncoKitDx	Molecular genetic screening of the *TP53* gene with amplicon-based NGS	48	CE IVD

ONCOLOGY _Liquid biopsy

Targeted Liquid Biopsy OncoKit

RUO

Tumor analysis using liquid biopsy.

Characteristics

- NGS panel and analysis software that test for the main biomarkers of clinical interest in non-small cell lung cancer and other solid tumors.
- Characterization of ctDNA from a liquid biopsy sample.
- Uses a single DNA sample to detect SNVs, indels, gene fusions, and CNVs.
- Bioinformatics analysis with the Data Genomics software and semiautomated generation of reports, including functional and clinical classification of the identified variants.
- Based on high sensitivity RNA-probe technology with UMI barcoding (unique molecular identifiers).
- Average coverage of 7300X and average coverage after UMI analysis of 2500X.
- The sensitivity, specificity, and repeatability of this test are greater than 99%. Reproducibility is >97%. Coverage (at a depth >100X) and uniformity of coverage of bases covered at >20X is >96%.
- Limit of detection: SNVs, indels, and structural variants: 1%; CNVs: gains of a total of 2.9 copies
- Compliant with quality specifications by the ISO 9001 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked analysis software.

Technical specifications

- Compatible sequencing platforms: Illumina NextSeq500 and NextSeq550Dx System
- Number of reactions: 24
- Number of samples per run: 8 samples with a 150-cycle NextSeq HIGH cartridge
- Sequencing: Paired-end (2 x 75 cycles).
- Sample type: Circulating cell-free DNA from a liquid biopsy sample
- Amount of input DNA: 10-50 ng
- Fully automated panel for Magnis NGS Prep System Dx equipment.
- CNV analysis requires that a positive control (REF: IMG-368) be used at least once.

Panel composition

- Sequencing of the coding regions of 13 genes:

ALK	*EGFR*	*KIT*	*MAP2K1*	*NRAS*	*RB1*	*ROS1*	*TP53*
BRAF	*ERBB2*	*KRAS*	*MET*	*PIK3CA*			

- Sequencing of 7 hotpost regions in the *NTRK1* and *NTRK3* genes.
- Capture of 8 fusion genes with any possible rearrangement (including previously reported intronic breakpoint regions):

ALK	*EGFR*	*ETV6*	*NRG1*	*NTRK1*	*NTRK2*	*RET*	*ROS1*

- Detection of CNVs throughout the whole genome. Validation of results and detection of copy-neutral LOH across the genome with a low-density SNP array.

ONCOHEMATOLOGY

Haematology OncoKitDx

NGS kit for the screening for the main molecular biomarkers associated with ALL, AML, CML, MDS, MPN, and other hematologic neoplasms).

Characteristics

- NGS panel for the study of the the following oncohematological diseases: acute myeloid leukemia (AML), chronic myeloid leukemia (CML), myeloproliferative neoplasms (MPN), acute lymphoid leukemia (ALL), and myelodysplastic syndromes.
- Uses a single DNA sample to detect SNVs, indels, gene fusions, CNVs, and pharmacogenetic variants.
- Bioinformatics analysis with the Data Genomics software and semiautomated generation of reports, including functional and clinical classification of the identified variants.
- STIDs can be included: Integrated sample identification and tracking system.
- Based on high sensitivity RNA-probe technology with UMI barcoding (unique molecular identifiers).
- Coverage (at depth >100x), specificity, sensitivity, repeatability, and reproducibility of this test are greater than 99%. The uniformity of bases covered at >20x is 98.4%.
- Limit of detection: SNVs and Indels: 2%, CNVs: 20% for copy losses and 10% for copy gains
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked kit and analysis software.

Technical specifications

- Compatible sequencing platforms: Illumina NextSeq 500/550/550Dx.
- Number of reactions: 24.
- Number of samples per run: 8 samples with a 150-cycle NextSeq MID cartridge.
- Sequencing: Paired-end (2 x 75 cycles).
- Sample type: DNA from peripheral blood and bone marrow.
- Amount of input DNA: 50-100 ng.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**
- CNV analysis requires that a positive control (REF: IMG-368) be used at least once.

Panel composition

IMG-363 Haematology OncoKitDx

- Sequencing of the coding regions of **76 genes**:

ARID5B	*CBL*	*DDX3X*	*GATA1*	*JAK2*	*NOTCH1*	*PTEN*	*SMC1A*	*TYK2*
ASXL1	*CDKN2A*	*DDX41*	*GATA2 (+I4)*	*JAK3*	*NPM1*	*PTK2B*	*SMC3*	*U2AF1*
ASXL2	*CDKN2B*	*DNMT3A*	*GATA3*	*KIT*	*NR3C1*	*PTPN11*	*SRP72*	*WT1*
ATRX	*CEBPA*	*EP300*	*HAVCR2*	*KMT2A*	*NRAS*	*RAD21*	*SRSF2*	*ZRSR2*
BCOR	*CHIC2*	*ETNK1*	*IDH1*	*KMT2C*	*P2RY8*	*RB1*	*STAG1*	
BCORL1	*CREBBP*	*ETV6*	*IDH2*	*KRAS*	*PAX5*	*RUNX1*	*STAG2*	
BLNK	*CSF3R*	*EZH2*	*IKZF1*	*MPL*	*PHF6*	*SETBP1*	*STAT5B*	
BRAF	*CSNK1A1*	*FBXW7*	*IL7R*	*NF1*	*PIGA*	*SF3B1*	*TET2*	
CALR	*CUX1*	*FLT3*	*JAK1*	*NFE2*	*PPM1D*	*SH2B3*	*TP53*	

- Capture of 27 fusion genes with any possible rearrangement (including previously reported intronic breakpoint regions):

ABL1	*CBFA2T3*	*EPOR*	*FUS*	*MEF2D*	*NPM1*	*PDGFRA*	*RBM15*	*STIL*
ABL2	*CBFB*	*ETV6*	*JAK2*	*MNX1*	*NUP214*	*PDGFRB*	*RUNX1*	*TAL1*
BCR	*CSF1R*	*FGFR1*	*KMT2A*	*MYH11*	*NUP98*	*RARA*	*SET*	*TCF3*

- Detection of CNVs throughout the whole genome (detection of hypo- and hyperploidy) or in an entire gene included in the panel. Validation of results and detection of copy-neutral LOH across the genome with a low-density SNP array.
- Detection of pharmacogenetic variants in ***ABCB1, CEP72, CYP2C9, ITPA, MTHFR, MTRR, NUTD15, PNPLA3, SLCO1B1,*** and ***TPMT.***

Chimerisms

Health in Code launches its **Imegen® Quimera qPCR and Imegen® Quimera dPCR kits**, the first CE-IVD-marked tests for the identification and relative (via real-time PCR) or absolute quantification (via digital PCR) of hematopoietic chimerism after allogeneic stem cell transplantation.

Chimerism analysis with **Imegen® Quimera dPCR kits** for digital PCR provides maximum sensitivity and precision to detect relapse early and, therefore, offer earlier medical action and better response to therapy.

Early prognosis with maximum accuracy.

Screening (qPCR, presence / absence)

Imegen-Quimera Screening Multiplex I and **Imegen-Quimera Screening Multiplex II are used for** multiplex real-time PCR genotyping of 34 different insertion/deletion (INDEL) polymorphisms to detect an informative marker for the follow-up of transplanted patients with **Imegen® Quimera qPCR kits**. The overall cumulative informativeness of both kits, along with SRY and RhD polymorphisims, is 99.96%.

Imegen-Quimera Screening Multiplex Plus and Imegen-Quimera Screening Multiplex Plus II are used for multiplex real-time PCR genotyping of 33 different insertion/deletion (INDEL) polymorphisms to detect an informative marker for the follow-up of transplanted patients with **Imegen® Quimera dPCR kits**. The overall cumulative informativeness of this panel, along with SRY and RhD polymorphisims, is 99.98%.

All Imegen-Quimera Screening kits were designed for real-time PCR on 7500 FAST Real-Time PCR System and StepOne Plus Real-Time PCR System (Applied Biosystems).

Monitoring (qPCR, dPCR, quantification)

Chimerism analysis by real-time PCR with the **Imegen® Chimera qPCR kit** performs relative quantification of hematopoietic chimerism in the post-transplant period in each assay, considering the amplification efficiency and the "cycle threshold" (Ct) values obtained for an endogenous control or calibrator, which is included in this kit as a positive control.

Compatible with 7500 FAST Real-Time PCR System and StepOne Plus Real-Time PCR System (Applied Biosystems).

Chimerism analysis of by digital PCR with **Imegen® Quimera dPCR kits** performs absolute quantification of hematopoietic chimerism in the post-transplant period, using the endogenous reference gene (beta-globin).

Compatible with QuantStudio 3D Digital PCR System and Absolute Q Digital PCR System (Applied Biosystem) and QX200™ Droplet Digital™ PCR System (Bio Rad) dPCR kits.

What are the **advantages** of Imegen-Quimera Screening Multiplex Plus and Imegen-Quimera dPCR kits?

- The screening allows 5 donor-recipient pair analyses.
- Absolute quantification (copy numbers).
- Endpoint detection.
- No need for technical replicates (dPCR performs 20,000 sample partitions).
- No need for standard curves or pre-transplant sample analysis.
- Small amounts of DNA required.
- Screening with accumulated information [screening + SRY + RhD] of 99.98%.
- Accurate results for increasing mixed chimerism (iMC) status.
- Excellent detection of residual cells (limit of quantification <0.05%).
- High assay reproducibility.
- Tracking software.
- Easy to perform and fast PCR procedures.
- Reduced hands-on time.

Imegen-Quimera qPCR kits

Reference	Kit name	Screening
IMG-116-18	Imegen-Quimera RhD	Additional marker
IMG-116-2	Imegen-Quimera SRY	Additional marker
IMG-116-10	Imegen-Quimera Q116-10I	Imegen-Quimera Screening Multiplex I IMG-116-24
IMG-116-11	Imegen-Quimera Q116-11I	
IMG-116-12	Imegen-Quimera Q116-12I	
IMG-116-13	Imegen-Quimera Q116-4D	
IMG-116-14	Imegen-Quimera Q116-5D	
IMG-116-17	Imegen-Quimera Q116-10D	
IMG-116-20	Imegen-Quimera Q116-20I	
IMG-116-21	Imegen-Quimera Q116-12D	
IMG-116-23	Imegen-Quimera Q116-23I	
IMG-116-3	Imegen-Quimera Q116-3I	
IMG-116-4	Imegen-Quimera Q116-4I	
IMG-116-5	Imegen-Quimera Q116-5I	
IMG-116-6	Imegen-Quimera Q116-6I	
IMG-116-7	Imegen-Quimera Q116-7I	
IMG-116-8	Imegen-Quimera Q116-8I	
IMG-116-9	Imegen-Quimera Q116-9I	

Reference	Kit name	Screening
IMG-116-16	Imegen-Quimera Q116-33I	Imegen-Quimera Screening Multiplex II IMG-116-74
IMG-116-66	Imegen-Quimera Q116-46I	
IMG-116-70	Imegen-Quimera Q116-50I	
IMG-116-73	Imegen-Quimera Q116-29D	
IMG-116-75	Imegen-Quimera Q116-37I	
IMG-116-76	Imegen-Quimera Q116-38I	
IMG-116-77	Imegen-Quimera Q116-44I	
IMG-116-78	Imegen-Quimera Q116-43I	
IMG-116-79	Imegen-Quimera Q116-49I	
IMG-116-80	Imegen-Quimera Q116-39I	
IMG-116-81	Imegen-Quimera Q116-47I	
IMG-116-82	Imegen-Quimera Q116-32I	
IMG-116-83	Imegen-Quimera Q116-31I	
IMG-116-84	Imegen-Quimera Q116-30D	
IMG-116-87	Imegen-Quimera Q116-24I	
IMG-116-88	Imegen-Quimera Q116-27D	

Reference	Kit name	Screening
IMG-116-27 Dry	Imegen-Quimera SRY dPCR Dry	Additional marker
IMG-116-29 Dry	Imegen-Quimera Q116-3l dPCR Dry	
IMG-116-30 Dry	Imegen-Quimera Q116-4l dPCR Dry	
IMG-116-35 Dry	Imegen-Quimera Q116-10l dPCR Dry	
IMG-116-34 Dry	Imegen-Quimera Q116-11l dPCR Dry	
IMG-116-36 Dry	Imegen-Quimera Q116-12D dPCR Dry	
IMG-116-31 Dry	Imegen-Quimera Q116-5l dPCR Dry	
IMG-116-37 Dry	Imegen-Quimera Q116-23l dPCR Dry	
IMG-116-38 Dry	Imegen-Quimera Q116-24l dPCR Dry	Imegen-Quimera Screening Multiplex Plus IMG-116-26
IMG-116-41 Dry	Imegen-Quimera Q116-27D dPCR Dry	
IMG-116-42 Dry	Imegen-Quimera Q116-28l dPCR Dry	
IMG-116-43 Dry	Imegen-Quimera Q116-29D dPCR Dry	
IMG-116-44 Dry	Imegen-Quimera Q116-30D dPCR Dry	
IMG-116-45 Dry	Imegen-Quimera Q116-31l dPCR Dry	
IMG-116-46 Dry	Imegen-Quimera Q116-32l dPCR Dry	
IMG-116-32 Dry	Imegen-Quimera Q116-6l dPCR Dry	
IMG-116-33 Dry	Imegen-Quimera Q116-7l dPCR Dry	

Reference	Kit name	Screening
IMG-116-50 Dry	Imegen-Quimera Q116-38l dPCR Dry	
IMG-116-51 Dry	Imegen-Quimera Q116-39l dPCR Dry	
IMG-116-40 Dry	Imegen-Quimera Q116-20l dPCR Dry	
IMG-116-28 Dry	Imegen-Quimera RhD dPCR Dry	
IMG-116-52 Dry	Imegen-Quimera Q116-41l dPCR Dry	
IMG-116-53 Dry	Imegen-Quimera Q116-42l dPCR Dry	
IMG-116-54 Dry	Imegen-Quimera Q116-43l dPCR Dry	
IMG-116-55 Dry	Imegen-Quimera Q116-44l dPCR Dry	
IMG-116-56 Dry	Imegen-Quimera Q116-45l dPCR Dry	Imegen-Quimera Screening Multiplex Plus II IMG-116-25
IMG-116-60 Dry	Imegen-Quimera Q116-46l dPCR Dry	
IMG-116-57 Dry	Imegen-Quimera Q116-47l dPCR Dry	
IMG-116-58 Dry	Imegen-Quimera Q116-49l dPCR Dry	
IMG-116-40 Dry	Imegen-Quimera Q116-20l dPCR Dry	
IMG-116-59 Dry	Imegen-Quimera Q116-50l dPCR Dry	
IMG-116-47 Dry	Imegen-Quimera Q116-33l dPCR Dry	
IMG-116-49 Dry	Imegen-Quimera Q116-37l dPCR Dry	
IMG-116-48 Dry	Imegen-Quimera Q116-9l dPCR Dry	

*Note: Does not include amplification polymerase. Supplied by the manufacturer of the thermal cycler.

Other hematology kits

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-108	Imegen-BCR-ABL Screening	Detection of the BCR-ABL t(9;22) (q34;q11) rearrangement	48 x 2	CE IVD
Real-Time PCR	IMG-109	Imegen-Inv16	Detection of the CBFB-MYH11 inv(16) (p13;q22) rearrangement	48 x 2	RUO
Real-Time PCR	IMG-111	Imegen-PML-RARA	Quantification of the PML-RARA t(15;17)(q22;q21) rearrangement	48 x 2	RUO
Real-Time PCR	IMG-121	Imegen-M-BCR-ABL	Quantification of the M-bcr:M-BCR-ABL p210 rearrangement	48 x 2	CE IVD
Real-Time PCR	IMG-122	Imegen-m-BCR-ABL	Quantification of the m-bcr:m-BCR-ABL p190 rearrangement	48 x 2	CE IVD
Real-Time PCR	IMG-130	Imegen-PML-RARA Screening	Detection of the PML-RARA t(15;17)(q22;q21) rearrangement	48 x 2	CE IVD
Real-Time PCR	IMG-235	Imegen-NPM1	Detection of mutations types A, B, and D in the *NPM1* gene	24	CE IVD
Real-Time PCR	IMG-236	Imegen-MPL	Detection of *MPL* W515L and W515K	24 x 2	CE IVD
Análisis de fragmentos (PCR)	IMG-237	Imegen-CALR	Detection of INDELs in the *CALR* gene	33	CE IVD
Análisis de fragmentos (PCR)	IMG-238	Imegen-FLT3	Detection of ITD mutations in the *FLT3* gene	33	CE IVD
Next-Generation Sequencing	IMG-317	TP53 OncoKitDx	Molecular genetic analysis of the *TP53* gene with amplicon-based NGS	48	CE IVD
Digital PCR	IMG-405	FLT3-TKD2 dPCR OncoKit	Detection and quantification of the variant D816V in the *KIT* gene	24	RUO
Digital PCR	IMG-407	cKit dPCR OncoKit	Detection and quantification of point mutations in the D835 and I836 codons of the TKD2 domain of *FLT3*	24	RUO

Other hematology kits

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-199	Imegen-Cambridge II	Genotyping of *SERPINC1* p.Ala368Ser, antithrombin	48	RUO
Real-Time PCR	IMG-212	Imegen-MTHFR	Genotyping of *MTHFR* c.677C>T	48	CE IVD
Real-Time PCR	IMG-214	Imegen-Factor II	Genotyping of *F2* c.20110G>A	48	CE IVD
Real-Time PCR	IMG-215	Imegen-Factor XII	Genotyping of *F12* c.46C>T	48	CE IVD
Real-Time PCR	IMG-216	Imegen-MTHFRII	Genotyping of *MTHFR* c.1298A>C	48	CE IVD
Real-Time PCR	IMG-217	Imegen-Factor V	Genotyping of *F5* p.Arg506Gln, Leiden	48	CE IVD
Real-Time PCR	IMG-218	Imegen-HFE	Genotyping of *HFE* p.Cys282Tyr, p.His63Asp, and p.Ser65Cys	48 x 3	CE IVD
Digital PCR	IMG-331	Imegen-Factor II dPCR	Genotyping of *F2* c.*97G>A (20210G>A), *prothrombin*	48	RUO
Digital PCR	IMG-332	Imegen-Factor V dPCR	Genotyping of *F5* c.1601G>A; p.Arg534Gln (R506Q), Leiden	48	RUO

MICROBIOLOGY

Neumo ViralDx

Imegen Neumo ViralDx kits test for the presence of the SARS-CoV-2 coronavirus and influenza A and B viruses as well as respiratory syncytial virus (RSV-A and RSV-B) in respiratory samples through the detection of specific genes and an endogenous human gene by real-time reverse transcription PCR (rRT-PCR).

They are based on real-time reverse transcription PCR aimed at the qualitative detection of SARS-CoV-2 RNA extracted from nasal swab, nasopharyngeal swab, and oropharyngeal swab samples taken by a medical specialist from the upper respiratory tract of individuals suspected of having COVID-19.

Technique	Reference	Kit name	Description	RXNS
Real-Time PCR	IMG-386	Imegen Neumo ViralDx	Multiplexed detection of the respiratory viruses SARS-CoV-2, Influenza A, Influenza B, and RSV, and of the endogenous human *RNaseP* gene	96

Technical characteristics

- Multiplexed qualitative assay that allows determining the presence of two SARS-CoV-2 targets, including the *ORF1ab and S* genes, and specific targets for influenza A and B viruses and for respiratory syncytial virus (RSV).
- The endogenous human gene, *RNaseP*, allows assessing the respiratory sample collection process, the quality of the sample, and the nucleic acid extraction process.
- Limit of detection: LoD <50 copies for the analyzed human viruses (FDA acceptance criterion)
- Cross-reactivity: diagnostic evaluation with pathological human samples confirm kit specificity against human enterovirus, bocavirus, adenovirus, rhinovirus, and metapneumovirus.
- Genomic inclusivity: *in silico* design based on 4,115 SARS-CoV-2 genomes (GISAID database).
- Detection of viral sequences of SARS-CoV-2, influenza, and RSV reported between 2016 and 2020.

Technical specifications

- Sample type: Purified RNA from human respiratory samples.
- Necessary sample volume: 10 µL.
- Number of reactions per sample: 1.
- Number of targets: 6.
- Fluorophores: FAM, VIC (HEX), Cy5, and TexasRed (ROX).
- Duration of the rRT-PCR program: 1h 20 min.
- Compatibles platforms: QuantStudio 5 Real-time PCR System (Applied Biosystems), 7500 FAST Real-time PCR System (Applied Biosystems) and CFX96 Real-time PCR System (BioRad).

SARS-COV-2

Imegen-SARS-CoV-2 kits test for the presence of the SARS-CoV-2 coronavirus in respiratory samples through the detection of specific genes and an endogenous human gene by real-time reverse transcription PCR (rRT-PCR).

They are based on real-time reverse transcription PCR aimed at the qualitative detection of SARS-CoV-2 RNA extracted from nasal swab, nasopharyngeal swab, oropharyngeal swab, and saliva samples taken by a medical specialist from the upper respiratory tract of individuals suspected of having COVID-19.

Technical characteristics

- Qualitative assay that allows determining the presence of three specific SARS-CoV-2 targets and a human endogenous target through the study of two multiplexed rRT-PCR reactions.
- The endogenous human gene, *RNaseP*, allows assessing the respiratory sample collection process, the quality of the sample, and the nucleic acid extraction process.
- Genomic inclusivity: *in silico* design based on 3,123 genomes (GISAID database).
- Approved by the Carlos III Health Institute (ISCiii).
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.

Technique	Reference	Kit name	Description	RXNS
Real-Time PCR	IMG-356	Imegen SARS-CoV-2	SARS-CoV-2 coronavirus testing by detection of the *N*, *S*, and *RdRp* genes and the endogenous human *RNaseP* gene	96

Technical specifications

- Sample type: Purified RNA from human respiratory samples.
- Necessary sample volume: 12 µL.
- Number of reactions per sample: 2.
- Number of targets: 4.
- Fluorophores: FAM, VIC (HEX).
- Duration of the rRT-PCR program: 1h 20 min.
- Limit of detection: 10 copies (FDA acceptance criterion).
- Compatibles platforms: 7500 FAST Real-Time PCR System (Applied Biosystems), StepOne Real-Time PCR System (Applied Biosystems), QuantStudio5 Real-Time PCR System (Applied Biosystems), CFX96 Touch Real-Time PCR System (BioRad), CFX96 Real-Time PCR System (BioRad), and Light Cycler 480 Real-Time PCR System (Roche).

Technique	Reference	Kit name	Description	RXNS
Real-Time PCR	IMG-376	Imegen SARS-CoV-2 Plus RNaseP	Multiplexed detection of the SARS-CoV-2 coronavirus *S*, *ORF1ab*, and *E* genes and the endougenous human *RNaseP* gene.	96

Technical specifications

- Sample type: Purified RNA from human respiratory samples.
- Necessary sample volume: 6 µL.
- Number of reactions per sample: 1.
- Number of targets: 4.
- Fluorophores: FAM, VIC (HEX), Cy5, and TexasRed (ROX).
- Duration of the rRT-PCR program: 1h 20 min.
- Limit of detection: 20 copies (FDA acceptance criterion).
- Compatibles platforms: 7500 FAST Real-Time PCR System (Applied Biosystems) and QuantStudio5 Real-Time PCR System (Applied Biosystems).

PHARMACOGENETICS

Action PharmaKitDx

NGS testing for the main molecular markers related to drug response described in pharmacogenomic clinical practice guidelines and technical data sheets of drug regulatory authorities.

Characteristics

- Dynamic kit whose target genes are updated as new scientific evidence becomes available.
- SNV and indel detection.
- Detection of CNVs in *CYP2D6*.
- Bioinformatic analysis through Client Site software
- STID: Integrated sample identification and tracking system.
- Average coverage: >900X, with 99.62% of bases covered at 30X.
- Sensitivity: >99%.
- Specificity: >99%.
- Repeatability: >99%.
- Reproducibility: >99%.
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked kit and analysis software.

Technical characteristics

- Compatible sequencing platforms: Illumina MiSeq System
- Number of reactions: 16.
- Number of samples per run: Illumina MiSeq V2 (300 cycles): 8.
- Sequencing: Paired-end (2 x 150 cycles).
- Sample type: DNA from peripheral blood.
- Amount of DNA per sample: 50-100 ng.
- Analyzed genomic region: 0.408 Mb.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**
- CNV analysis requires that a positive control (REF: IMG-417) be used at least once.

IMG-401 Action PharmaKitDx [335 genes]

ABCB1
ABCB11
ABCB4
ABCB7
ABCC1
ABCC2
ABCC3
ABCC4
ABCC5
ABCC6
ABCC8
ABCC9
ABCG1
ABCG2
ACE
ACKR1
ACYP2
ADA
ADD1
ADGRB3
ADGRL2
ADH1A
ADH1B
ADH1C
ADORA2A
ADRB1
ADRB2
AFF3
AHR
AKR1E2
ALDH1A1
ALDH2
ALOX5
ALPL
AMPD1
ANKK1
APOA5
APOE
ARL14
ATIC
ATP7A
BDNF
BMP2
BMP7
BRINP1
BTG1
C11orf65
C1orf167
C8orf34
CACNA1S
CADM2
CALU
CARD8
CBR3
CCDC179
CCHCR1
CCR5
CD2
CD226
CD69
CD80
CD84
CD86
CDA
CEP72
CES1
CES2
CETP
CHRNA3
CHRNB2
CHST7
CHUK
CLEC2D
CLSTN2
CNTN5
COL22A1
COMT
COQ2
CREB1
CRHR1
CRHR2
CRP
CSMD1
CSMD3
CUL1
CYP17A1
CYP19A1
CYP1A1
CYP1A2
CYP1B1
CYP2A13
CYP2A6
CYP2B6
CYP2C18
CYP2C19
CYP2C8
CYP2C9
CYP2D6
CYP2E1
CYP2F1
CYP2J2
CYP2S1
CYP3A4
CYP3A43
CYP3A5
CYP3A7
CYP4B1
CYP4F2
DCK
DHFR
DHODH
DLG2
DOCK1
DPP6
DPYD
DRD1
DRD2
DRD3
DRD4
DYNC2H1
EGF
EGLN3
EMCN
ENOX1
EPHX1
ERCC1
ESR1
ESR2
ETV1
EYA4
F5
FAAH
FAR1
FCGR2A
FCGR2B
FCGR3A
FDPS
FKBP5
FLT3
FMO2
FMO3
FOXP3
FPGS
FTO
G6PD
GABBR2
GALNT18
GATM
GFRA1
GFRA2
GGCX
GGH
GNB3
GP1BA
GRIK4
GSTM1
GSTP1
GSTT1
HAS3
HLA-A
HLA-B
HLA-C
HLA-DRB1
HLA-E
HLA-G
HMGCR
HTR1A
HTR2A
HTR2C
HTR3A
IFNG
IFNGR2
IFNL3
IFNL4
IGHM
IKBKB
IL10
IL12B
IL17A
IL17F
IL18
IL1B
IL1R1
IL2
IL4R
IL6
IL6R
IRAK3
ITGB7
ITPA
KCNE1
KCNH2
KCNIP1
KCNJ11
KIAA0391
KIF6
KLRC1
KLRD1
LMO3
LMX1A
LPA
LRPAP1
LRRC55
LTC4S
MAP2K6
MAP3K1
MAP3K14
MAP3K7
MAPK14
MAPKAPK2
MAPT
MC4R
MED15
MICA
MMP20
MRPL36
MS4A1
MTHFD1
MTHFR
MTR
MTRR
MYD88
NAT1
NAT2
NAV2
NEBL
NEDD4L
NFIB
NFKBIA
NFKBIB
NFKBIE
NLRP3
NQO1
NR1I2
NR2F2
NR3C1
NT5C2
NTRK2
NUBPL
NUDT15
OPRD1
OPRM1
OR2B11
OSR1
P2RY1
P2RY12
PDZD2
PLA2G4A
PNPLA3
PRELID3B
PRKCA
PRSS53
PTGFR
PTGIS
PTGS1
PTGS2
PTPRC
PTPRM
QKI
RAD51B
RHOBTB1
RPS6KA5
RYR1
SCN1A
SCN5A
SCNN1A
SEMA3C
SHMT1
SLC15A1
SLC15A2
SLC16A7
SLC19A1
SLC22A1
SLC22A11
SLC22A2
SLC22A6
SLC28A3
SLC46A1
SLC6A4
SLC9A7
SLCO1A2
SLCO1B1
SLCO1B3
SLCO1C1
SLCO2B1
SOD2
SP3
SRY
STAT4
STRBP
SULT1A1
SULT1A2
SYNM
TANC1
TBXAS1
TCF7L2
TEC
TGFB1
TGFBR2
TLR1
TLR10
TLR2
TLR4
TLR5
TNF
TNFAIP3
TNFRSF1A
TNFRSF1B
TNFSF13B
TP53
TPMT
TRAF1
TUBB2A
TYMS
UGT1A1
UGT1A10
UGT1A3
UGT1A4
UGT1A5
UGT1A6
UGT1A7
UGT1A8
UGT1A9
UGT2B10
UGT2B15
UGT2B17
UGT2B7
UMPS
VDR
VKORC1
WDR27
XPC
XRCC1
YEATS4
ZNF595

Target genes

DPYD PharmaKitDx

Qualitative analysis kit for pharmacogenetics related to cancer treatment studies based on the detection of informative polymorphisms in the *DPYD* gene (DPYD*2A, DPYD*13, DPYD*hapB3, and D494V).

IMG-413 DPYD PharmaKitDx	4x48 rxn

Characteristics

- **Qualitative analysis that can detect the following** *DPYD* alleles:
  - *DPYD**2A [NM_000110.4: c.1905+1G>A (rs3918290)]
  - *DPYD**13 [NM_000110.4: c.1679T>G (rs55886062)]
  - *DPYD**hapB3 [NM_000110.4: c.1129-5923C>G (rs75017182)]
  - *DPYD*-D949V [NM_000110.4: c.2846A>T (rs67376798)]
- Pharmacogenetic analysis of response to fluoropyrimidine-based chemotherapy treatments, which include 5-fluorouracil (5-FU) and its prodrugs, capecitabine and tegafur, offering an optional method for dose adjustment in each case.
- High sensitivity and specificity: limit of detection set at 1 ng of genomic DNA.
- Includes a positive heterozygous control for each of the analyzed variants.
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.

Technical characteristics

- Recommended amount of DNA: 50 ng.
- Sample type: Peripheral blood (germline).
- Number of reactions per sample: 4.
- Number of targets: 4.
- Set-up and preparation: 15 min.
- Duration of the PCR program: 1h 30 min.
- Compatible platforms: StepOne Plus Real-Time PCR System (Applied Biosystems) and 7500 FAST Real-Time PCR System (Applied Biosystem).

IMMUNOLOGY

Immunology kits

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-289	Imegen HLA-B27	Detection of the HLA-B27 allele	48	CE IVD
Real-Time PCR	IMG-306	Imegen HLA-B57:01	Detection of the HLA-B*57:01 allele	48	CE IVD
Real-Time PCR	IMG-341	Imegen-Coeliac	Detection of alleles HLA-DQA1*05, HLA-DQA1*03, and HLA-DQB1*03:02 and allele discrimination between HLA-DQB1*02 and the rest of HLA-DQB1 alleles	48 x 3	CE IVD
Real-Time PCR	IMG-420	Coeliac InmunoKit	Study of susceptibility to developing celiac disease by analyzing the alleles HLA-DQA1*02, *03, *05 and HLA-DQB1*03:01 and *03:02, as well as allelic discrimination between HLA-DQB1*02 and all other HLA-DQB1 alleles	48 x 4	RUO

CARDIOLOGY

Inherited CardioKitDx

Analysis of genetic alterations related to sudden death or supraventriular arrhythmias (atrial fibrillation), channelopathies, and cardiac conduction diseases, through an automated laboratory process by NGS sequencing.

Characteristics

- **SNV and indel** analysis in genes with full exonic regions and intronic regions of interest, as well as **CNV** analysis.
- Bioinformatic analysis through Client Site software.
- STID: Integrated sample identification and tracking system.
- Coverage: 99.8% of bases covered at a minimum depth of 30X.
- Uniformity: 99.1% of bases covered at >20% of mean coverage.
- Sensitivity and specificity: >99.9%.
- Repeatability and reproducibility: >99%.
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing material.
- CE-IVD-marked kit and analysis software.

Technical specifications

- Compatible sequencing platforms: Illumina NextSeq500/550.
- Sample type: DNA from peripheral blood.
- Number of reactions: 16.
- Number of samples per run: 16 samples with 150-cycle NextSeq MID cartridge.
- Sequencing: Paired-end (2x75 cycles).
- Amount of input DNA: 200 ng.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**

IMG-390 Inherited CardioKitDx [261 genes]

A2ML1	*CALM1*	*DNAJC19*	*GJA5*	*KCNJ2*	*MTO1*	*PDLIM3*	*SCN1B*	*TBX5*
AARS2	*CALM2*	*DNM1L*	*GLA*	*KCNJ5*	*MYBPC3*	*PERP*	*SCN2B*	*TCAP*
ABCC9	*CALM3*	*DOLK*	*GLB1*	*KCNJ8*	*MYBPHL*	*PHKA1*	*SCN3B*	*TECRL*
ACAD9	*CALR*	*DSC2*	*GNB2*	*KCNK17*	*MYH11*	*PITX2*	*SCN4B*	*TGFB3*
ACADVL	*CALR3*	*DSG2*	*GNPTAB*	*KCNK3*	*MYH6*	*PKD2*	*SCN5A*	*TMEM175*
ACTA1	*CAPN3*	*DSP*	*GPD1L*	*KCNQ1*	*MYH7*	*PKP2*	*SCO2*	*TMEM43*
ACTC1	*CASQ2*	*DTNA*	*GREM2*	*KLF10*	*MYL2*	*PKP4*	*SDHA*	*TMEM70*
ACTN2	*CASZ1*	*ELAC2*	*GSK3B*	*KLHL24*	*MYL3*	*PLN*	*SGCA*	*TMOD1*
AGK	*CAV3*	*EMD*	*GUSB*	*KRAS*	*MYLK2*	*PMM2*	*SGCB*	*TNNC1*
AGL	*CAVIN1*	*EYA4*	*GYG1*	*LAMA2*	*MYOM1*	*PPA2*	*SGCD*	*TNNI3*
AGPAT2	*CAVIN4*	*FAH*	*HCN4*	*LAMA4*	*MYOT*	*PPCS*	*SGCG*	*TNNI3K*
AKAP9	*CBL*	*FBXO32*	*HFE*	*LAMP2*	*MYOZ2*	*PPP1CB*	*SHOC2*	*TNNT2*
AKT1	*CDH2*	*FGF12*	*HRAS*	*LDB3*	*MYPN*	*PPP1R13L*	*SLC22A5*	*TOR1AIP1*
ALMS1	*COA5*	*FHL1*	*IDH2*	*LDLR*	*NEBL*	*PRDM16*	*SLC25A3*	*TPM1*
ALPK3	*COA6*	*FHL2*	*ILK*	*LIAS*	*NEXN*	*PRKAG2*	*SLC25A4*	*TRDN*
ANK2	*COL7A1*	*FHOD3*	*IRX3*	*LMNA*	*NF1*	*PSEN1*	*SLMAP*	*TRIM54*
ANK3	*COQ2*	*FKRP*	*ISM2*	*LMOD2*	*NKX2-5*	*PSEN2*	*SNTA1*	*TRIM63*
ANKRD1	*COX15*	*FKTN*	*JARID2*	*LZTR1*	*NKX2-6*	*PTPN11*	*SOS1*	*TRPM4*
ANO5	*COX6B1*	*FLNC*	*JPH2*	*MAP2K1*	*NNT*	*QRSL1*	*SOS2*	*TSFM*
ATP5F1E	*CRYAB*	*FOXD4*	*JUP*	*MAP2K2*	*NONO*	*RAF1*	*SPEG*	*TTN*
ATPAF2	*CSNK1A1*	*FOXRED1*	*KAT6B*	*MAP3K8*	*NOS1AP*	*RANGRF*	*SPRED1*	*TTR*
BAG3	*CSRP3*	*FXN*	*KCNA5*	*MEF2C*	*NOTCH1*	*RASA1*	*SPRY1*	*TXNRD2*
BRAF	*CTNNA1*	*GAA*	*KCND2*	*MIB1*	*NPPA*	*RASA2*	*SRY*	*VCL*
BSCL2	*CTNNA3*	*GATA4*	*KCND3*	*MIR208A*	*NRAP*	*RBM20*	*SURF1*	*WISP1*
C10orf71	*CTNNB1*	*GATA5*	*KCNE1*	*MIR208B*	*NRAS*	*RBM24*	*SYNE1*	*WT1*
CACNA1C	*CHRM2*	*GATA6*	*KCNE2*	*MLYCD*	*OBSCN*	*RIT1*	*SYNE2*	*XK*
CACNA1D	*DES*	*GATAD1*	*KCNE3*	*MRPL3*	*OBSL1*	*RRAS*	*SYNGAP1*	*ZBTB17*
CACNA2D1	*DLD*	*GFM1*	*KCNE5*	*MRPL44*	*OPA3*	*RYR2*	*TAZ*	*ZFHX3*
CACNB2	*DMD*	*GJA1*	*KCNH2*	*MRPS22*	*PDHA1*	*SCN10A*	*TBX20*	*ZFPM2*

Target genes

In addition to coding regions and ± 50 bp of adjacent intronic regions, Inherited CardioKitDx includes:

- Extension to ±100 bp of the intronic regions of the *FBN1, PKP2, LDLR, FLNC, DSP,* and *LMNA* genes.
- The *MYBPC3* gene completely covered.
- Promotor and UTR regions in the *LMNA, TNNT2, SCN5A, PLN, MYH7, LDLR, TNNI3,* and *DMD* genes.
- For more sensitive CNV calling, equispaced intronic regions of *DMD* and *PKP2* have been covered.
- Regions of the microRNAs MIR208a and MIR208b, located in introns of *MYH6* and *MYH7*.

NEPHROLOGY

Inherited NephroKitDx

Kit with recognized diagnostic and prognostic value among the different kidney pathologies (glomerular diseases, tubulointerstitial diseases, kidney cystic diseases, hereditary cancer, CAKUT, and other syndromes with renal malformations), both in adults and in children, that analyzes the main genes associated with monogenic inherited nephropathies.

Characteristics

- **SNV and indel analysis** in genes with full exonic regions and intronic regions of interest, as well as CNV analysis.
- Bioinformatics analysis with the Data Genomics software.
- STIDs can be included: Integrated sample identification and tracking system.
- Average coverage without duplicates: 1200X.
- Coverage: 99.7% of bases covered at 50X.
- Uniformity: 97.5% of bases covered at >20% of mean coverage.
- Sensitivity: >99%.
- Specificity: >99%.
- Repeatability: >99%.
- Reproducibility: >99%.
- Compliant with quality specifications by the ISO 13485 and ISO 14001 standards regarding manufacturing materials.
- CE-IVD-marked and analysis software.

Technical specifications

- Compatible sequencing platforms: Illumina NextSeq 500 System and Illumina NextSeq 550Dx System.
- Number of reactions: 16.
- Number of samples per run: 8 samples in a 300-cycle MID cartridge.
- Sequencing: Paired-end (2x150 cycles).
- Sample type: DNA from peripheral blood.
- Amount of input DNA: 50-100 ng.
- **Fully automated panel for Magnis NGS Prep System Dx equipment.**

IMG-370 Inherited NephroKitDx [529 genes]

ACE, ACTB, ACTG1, ACTN4, ADAMTS13, ADAMTS9, ADCY10, AGT, AGTR1, AGXT, AHI1, ALG1, ALG8, ALG9, ALMS1, ALPL, AMER1, ANKFY1, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOA4, APOE, APOL1, APRT, AQP2, ARHGAP24, ARHGDIA, ARL13B, ARL3, ARL6, ARMC9, ATN1, ATP6V0A4, ATP6V1B1, ATP6V1C2, AVIL, AVP, AVPR2, B2M, B3GLCT, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BMP2, BMP4, BMPER, BNC2, BSCL2, BSND, C1QA, C1QB, C1QC, C3, C4A, C4B, C8ORF37, CA1, CA2, CASP10, CASR, CC2D2A, CCBE1, CCDC28B, CCL2, CCNQ, CD151, CD2AP, CD46, CD81, CD96, CDC42, CDC5L, CDC73, CDK10, CDK20, CDKN1C, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP55, CEP83, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CFP, CHD1L, CHD7, CHRM3, CISD2, CIT, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COPA, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CPLANE1, CPT1A, CRB2, CRKL, CSPP1, CTH, CTLA4, CTNS, CTU2, CUBN, CUL3, CYP24A1, DACH1, DACT1, DCDC2, DCHS1, DDX59, DGKE, DHCR7, DLC1, DMP1, DNAJB11, DNASE1, DSTYK, DVL1, DVL3, DYNC2H1, DYNC2LI1, DZIP1L, EGF, EHHADH, EMP2, ENPP1, EP300, ESCO2, ETFA, ETFB, ETFDH, ETV4, EXOC8, EYA1, FAH, FAM20A, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCL, FAS, FASLG, FAT1, FAT4, FCGR2A, FCGR3A, FGA, FGF10, FGF20, FGF23, FGFR2, FGFR3, FLCN, FLNA, FMN1, FN1, FOXI1, FOXP1, FRAS1, FREM1, FREM2, FUZ, FXYD2, G6PD, GANAB, GAPVD1, GATA3, GCM2, GDF11, GEMIN4, GLA, GLI3, GLIS2, GLIS3, GNA11, GPC3, GPHN, GREB1L, GRHPR, GRIP1, H19, HAAO, HAS2, HES7, HGD, HNF1A, HNF1B, HNF4A, HOGA1, HPRT1, HPSE2, HSD11B2, HSD17B4, HSPA9, IFT122, IFT140, IFT172, IFT27, IFT43, IFT46, IFT52, IFT74, IFT80, IFT81, INF2, INPP5E, INTU, INVS, IQCB1, IRF5, ITGA3, ITGA8, ITGAM, ITGB4, ITSN1, ITSN2, JAG1, JAM3, KANK1, KANK2, KANK4, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ15, KCNJ16, KCTD1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, KLHL3, KMT2D, KYNU, LAGE3, LAMA5, LAMB2, LCAT, LMNA, LMX1B, LRIG2, LRP2, LRP4, LRP5, LYZ, LZTFL1, MAD2L2, MAFB, MAGED2, MAGI2, MAPKBP1, MBTPS2, MCM5, MKKS, MKS1, MMACHC, MNX1, MOCOS, MOCS1, MUC1, MYCN, MYH9, MYO1E, NAA10, NCAPG2, NDUFAF3, NDUFB8, NDUFS2, NEK1, NEK8, NEU1, NFIA, NIPBL, NOS1AP, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR3C2, NRIP1, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, NXN, OCRL, OFD1, OPLAH, OSGEP, PAX2, PAX8, PBX1, PCBD1, PCSK5, PDE6D, PDSS1, PDSS2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGM3, PHEX, PHGDH, PIBF1, PIEZO2, PIGN, PIGT, PKD1, PKD2, PKHD1, PLA2R1, PLCE1, PMM2, PODXL, PORCN, PRKCD, PRKCSH, PRPS1, PTEN, PTPN22, PTPRO, PUF60, RAD21, RAI1, RARRES1, REN, RERE, RET, RFWD3, RMND1, RNU4ATAC, ROBO2, ROR2, RPGRIP1L, RPL26, SALL1, SALL4, SARS2, SCARB2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC61B, SEC63, SEMA3E, SF3B4, SGPL1, SI, SIX1, SIX2, SIX5, SLC12A1, SLC12A2, SLC12A3, SLC12A7, SLC22A12, SLC26A1, SLC26A7, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC3A1, SLC41A1, SLC4A1, SLC4A2, SLC4A4, SLC5A1, SLC5A2, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC9A3R1, SLIT2, SMARCAL1, SNRPB, SON, SOX11, SOX17, SPRY2, SRGAP1, STAT1, STAT4, STK11, STRA6, STS, SUFU, SYNPO, TBC1D24, TBC1D8B, TBX18, TBX4, TBXT, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, THOC6, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM260, TMEM67, TNFSF4, TNIP1, TNS2, TNXB, TP53RK, TP63, TPRKB, TRAF3IP1, TRAP1, TREX1, TRIM32, TRIP11, TRPC6, TRPM6, TRPS1, TRRAP, TSC1, TSC2, TTC21B, TTC37, TTC8, TXNDC15, TXNL4A, UMOD, UPK3A, USP8, VANGL1, VANGL2, VDR, VHL, VIPAS39, VPS33B, VTN, WDPCP, WDR19, WDR34, WDR35, WDR60, WDR72, WDR73, WFS1, WNK1, WNK4, WNT3, WNT4, WNT5A, WNT7A, WT1, XDH, XPNPEP3, XPO5, XRCC2, XRCC4, ZAP70, ZIC3, ZMPSTE24, ZNF148, ZNF365, ZNF423

Target genes

Inherited NephroKitDx also includes a total of 98 intronic variants of interest in the following 32 genes:
BBS1, BBS4, CD46, CEP290, CFH, CLCN7, COL4A5, CPLANE1, CPT1A, CUBN, FLCN, FLNA, GLA, HNF1A, IRF5, JAM3, OCRL, PHEX, PKHD1, PMM2, SDCCAG8, SLC12A3, SLC2A9, SNRPB, TMEM107, TSC1, TSC2, TXNL4A, UBA5, UMOD, VHL and *ZAP70*.

OTHER SYNDROMES

Neurology

Technique	Reference	Kit name	Description	RXNS	
Fragment analysis (PCR & TP-PCR)	IMG-152	Imegen-SCAs	Detection of repeat expansions in the *SCA1, SCA2, SCA3, SCA6, SCA7, SCA12, SCA17* and *DRPLA* genes	12 x 10	CE IVD
Fragment analysis (PCR & TP-PCR)	IMG-173	Imegen-DM1	Detection of the CTG repeat expansion in the *DMPK* gene	12 x 2	CE IVD
Fragment analysis (PCR & TP-PCR)	IMG-155	Imegen-Friedreich	Detection of GAA repeat expansion in the *FXN* gene	12 x 2	RUO
Fragment analysis (PCR & TP-PCR)	IMG-154	Imegen-Huntington	Detection of CAG repeat expansion in the *HTT* gene	12 x 2	CE IVD

Reproductive genetics

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-212	Imegen-MTHFR	Genotyping of the c.677C>T mutation in the *MTHFR* gene	48	CE IVD
Real-Time PCR	IMG-214	Imegen-Factor II	Genotyping of the c.20110G>A mutation in the *F2* (prothrombin) gene	48	CE IVD
Real-Time PCR	IMG-215	Imegen-Factor XII	Genotyping of the c.46C>T mutation in the *F12* gene	48	CE IVD
Real-Time PCR	IMG-216	Imegen-MTHFRII	Genotyping of the c.1298A>C mutation in the *MTHFR* gene	48	CE IVD
Real-Time PCR	IMG-217	Imegen-Factor V	Genotyping of the p.Arg506Gln mutation in the *F5* gene (Leiden)	48	CE IVD
Digital PCR	IMG-331	Imegen-Factor II dPCR	Genotyping of the c.*97G>A (20210G>A) mutation in *F2* (Prothrombin)	48	RUO
Digital PCR	IMG-332	Imegen-Factor V dPCR	Genotyping of the c.1601G>A; p.Arg534Gln (R506Q) mutation in *F5* (Leiden)	48	RUO

Pediatry

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-211	Imegen-Alfa-1-AT	Genotyping of p.Glu342Lys (PI-Z allele) and p.Glu246Val (PI-S allele) polymorphisms in the *SERPINA1* gene	48 x 2	CE IVD
Real-Time PCR	IMG-341	Imegen-Coeliac	Detection of alleles HLA-DQA1*05, HLA-DQA1*03, and HLA-DQB1*03:02 and allele discrimination between HLA-DQB1*02 and the rest of HLA-DQB1 alleles	48 x 3	CE IVD
Fragment analysis	IMG-288	Imegen-Gilbert Plus	Detection of the A(TA)7TAA, A(TA)5TAA. and A(TA)8TAA mutant alleles and the wild-type allele in the *UGT1A1* gene promotor	48	CE IVD
Real-Time PCR	IMG-420	Coeliac InmunoKit	Study of susceptibility to developing celiac disease by analyzing the alleles HLA-DQA1*02, *03, *05 and HLA-DQB1*03:01 and *03:02, as well as allelic discrimination between HLA-DQB1*02 and all other HLA-DQB1 alleles	48 x 4	RUO

Other digestive system kits

Technique	Reference	Kit name	Description	RXNS	
Real-Time PCR	IMG-211	Imegen-Alfa-1-AT	Genotyping of p.Glu342Lys (PI-Z allele) and p.Glu246Val (PI-S allele) polymorphisms in the *SERPINA1* gene	48 x 2	CE IVD
Real-Time PCR	IMG-341	Imegen-Coeliac	Detection of alleles HLA-DQA1*05, HLA-DQA1*03, and HLA-DQB1*03:02 and allele discrimination between HLA-DQB1*02 and the rest of HLA-DQB1 alleles	48 x 3	CE IVD
Fragment analysis (PCR & TP-PCR)	IMG-288	Imegen-Gilbert Plus	Detection of the A(TA)7TAA, A(TA)5TAA. and A(TA)8TAA mutant alleles and the wild-type allele in the *UGT1A1* gene promotor	48	CE IVD
Real-Time PCR	IMG-420	Coeliac InmunoKit	Study of susceptibility to developing celiac disease by analyzing the alleles HLA-DQA1*02, *03, *05 and HLA-DQB1*03:01 and *03:02, as well as allelic discrimination between HLA-DQB1*02 and all other HLA-DQB1 alleles	48 x 4	RUO

Associated components

Technique	Reference	Kit name	Description	RXNS	
Next-Generation Sequencing	IMG-327	96 Index Plate for Illumina	Plate with 96 indexes multiplexed with the universal index for sequencing on Illumina platforms. Compatible with the Hereditary OncoKitDx kit (without index).	96	RUO
Next-Generation Sequencing	IMG-234	Imegen-Sample Tracking A	Plasmids to track samples during genetic analysis with NGS. Compatible with amplicon-based NGS kits.	12 x 65 µL	RUO
Next-Generation Sequencing	IMG-311	Imegen-Sample Tracking B	Plasmids to track samples during genetic analysis with NGS. Compatible with amplicon-based NGS kits.	12 x 65 µL	RUO
Next-Generation Sequencing	IMG-340	Imegen-Sample Tracking components	Plasmids to track samples during genetic analysis with NGS. Compatible with capture-based NGS kits.	24 x 15 µL	RUO
Real-Time PCR	IMG-287	General Master Mix	Real-time PCR Master Mix HotStart 2x.	600 µL	RUO
Next-Generation Sequencing	IMG-368	imegen-gDNA Reference Samples	Male and female germline DNA for use as a control for CNV analysis in the following references: IMG-363, IMG-365, IMG-400, IMG-415, and IMG-419.	2 x 30 µl	RUO
Next-Generation Sequencing	IMG-417	imegen-gDNA Pharma Reference Samples	Germline DNA for use as a control for CNV analysis and subsequent haplotyping of Action PharmaKitDx.	30 µl	RUO
Next-Generation Sequencing	IMG-416	Enzymatic Fragmentation Components	Enzyme and buffer for enzymatic DNA fragmentation. Compatible with all enzymatic fragmentation and library preparation kits using the Magnis NGS Prep System.	24	RUO

Coming soon...

> Lymphomas B & T OncoKitDx

> NPM1 OncoKitDx dPCR

> BCR-ABL1 OncoKitDx dPCR

PRE-TEST AND POST-TEST COUNSELING

All of our studies offer pretest and posttest counseling

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