Av/ Diagonal 640, 6th floor
08017 Barcelona (Spain)
Tel.: +34 93 4453930
Fax: +34 93 2287899
VAT: ESB66343344

MOLECULAR TESTING OF TUMOR TISSUE SAMPLES

CODE	Recommended studies by pathology
CS-T-002	Lung tumors*. Gene sequencing including EGFR, KRAS, BRAF (NGS Somatic QIAseq DNA Custom Extended Panel-30 genes). nCounter RNA Fusion and Gene Expression Tumor Panel (including ALK, ROS, RET) IHQ PDL1
CS-T-004	Colorectal tumors* Gene sequencing including KRAS, BRAF, NRAS (NGS QIAseq DNA Custom-19 genes). IHQ Repair genes (MLH1, MSH2, MSH6, PMS2)
MS-T-002	Breast, gynecological, prostate tumors* Gene sequencing including BRCA1, BRCA2, (NGS QIAseq DNA Custom Breast_Gynecological Panel-11 genes)
CS-T-003	Urothelial tumors* Gene sequencing including FGFR2, FGFR3 (NGS Somatic QIAseq DNA Custom Extended Panel-30 genes). nCounter RNA Fusion Urothelial Carcinoma Panel
CS-T-001	Other solid tumors* (Melanoma, pancreas, gastric, etc.) Gene sequencing including EGFR, KRAS, BRAF, NRAS (NGS Somatic QIAseq DNA Custom Extended Panel-30 genes). nCounter RNA Fusion and Gene Expression Tumor Panel (including ALK, ROS, RET)

MULTIGENIC MOLECULAR PANELS

CODE	Study mutations (SNVs/indels) and copy number variation (CNVs)
MS-T-001	NGS Somatic QIAseq DNA Custom Panel (19 genes) Detection of somatic alterations in 19 genes with clinical relevance in cancer. Genes: ALK, BRAF, CDK4, CDK6, EGFR, ERBB2, ERBB4, FGFR1, IDH1, IDH2, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, ROS1, STK11, TP53. (Includes METΔ14 splicing variant)
MS-T-002	NGS Somatic QIAseq DNA custom Breast-Gynecological Panel (11 genes) Detection of somatic alterations in 11 genes with clinical relevance in breast, gynecological, and prostate cancer. Genes: AKT, BRCA1, BRCA2, CTNNB1, ERBB2, ESR1, PALB2, PIK3CA, POLE, PTEN, TP53
MS-T-003	NGS Somatic QIAseq DNA Custom Extended Panel (30 genes) Detection of somatic alterations in 30 genes with clinical relevance in cancer. Genes: ALK, ARID1A, BRAF, CDK4, CDK6, EGFR, ERBB2, ERBB4, FAT1, FGFR1, FGFR2, FGFR3, IDH1, IDH2, KEAP1, KIT, KRAS, MET, MYC, NFE2L2, NRAS, PDGFRA, PIK3CA, POLD1, POLE, RET, ROS1, SETD2, STK11, TP53. (Includes METΔ14 splicing variant)
MS-T-005	NGS Somatic QIAseq DNA Repair Genes Panel (28 genes) Detection of somatic alterations in 28 genes, including some involved in the DNA repair pathway. Genes: APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, CTNNB1, EPCAM, FANCA, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PIK3CA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53
CODE	**Study of fusion genes and gene expression signatures**
MS-T-006	nCounter RNA Fusion and Gene Expression Tumor Panel (12 genes)** Detection of somatic alterations in 12 genes with clinical relevance in cancer. Fusions: ALK, FGFR1, FGFR2, FGFR3, NRG1, NTRK1, NTRK2, NTRK3, RET, ROS1. Splicing variant: METΔ14, EGFRvIII. Gene expression analysis: ALK, ATM, BARD1, BID, BRCA1, BRCA2, CCNE1, CD274, CD4, CD8A, CDK2, EGFR, FGFR1, 2, 3, FOXP3, GZMM, IFNG, KRAS, NRG1, NTRK1, 2, 3, PDCD1, PTEN, RET, ROS1, STK11, TP53
MS-T-007	nCounter RNA Fusion Urothelial Carcinoma Panel (2 genes) Fusions: FGFR2, FGFR3. Identification of the variants: FGFR2-BICC1, FGFR2-CASP7, FGFR3ex17-TACC3ex10, FGFR3ex17-TACCex10, FGFR3-BAIAP2L1
MS-T-008	PROSIGNA Genetic prognostic signature for breast cancer (minimum two samples)

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INDIVIDUAL GENE MUTATIONS (TaqMan® allelic discrimination qPCR assay)

CODE	SERVICE
ISM-T-001	Single mutation detection (specify mutation)
ISM-T-002	EGFR exon 19 del and EGFR exon 21 (L858R, L861Q) mutations
ISM-T-003	EGFR exon 19 de, EGFR exon 21 (L858R, L861Q) and EGFR exon 20 T790M mutations
ISM-T-004	EGFR exon 20 resistance mutation T790M
ISM-T-005	EGFR exon 20 resistance mutation C797S
ISM-T-006	KRAS (codons 12, 13 and 61)
ISM-T-007	BRAF (V600E, V600K, V600R)
ISM-T-008	PIK3CA (codons 542, 545 and 1047)
ISM-T-009	ESR1 (codons 380, 536, 537, 538)

ANATOMICAL PATHOLOGY DIAGNOSIS

CODE	SERVICE
PP-T-003	Anatomical pathology diagnosis (H&E and 4 IHC analysis)
PP-T-004	Confirmation of anatomical pathology diagnosis (Previous AP report required)

MAIN FISH ASSAYS

CODE	SERVICE
ISF-T-001	HER-2/neu amplification**
ISF-T-002	ALK fusions**
ISF-T-003	ROS1 fusions
ISF-T-004	RET fusions
ISF-T-005	MET amplification
ISF-T-006	EGFR amplification
ISF-T-007	KRAS amplification
ISF-T-008	NTRK1 fusions
ISF-T-009	NTRK2 fusions
ISF-T-010	NTRK3 fusions
ISF-T-011	FGFR1 fusions
ISF-T-012	FGFR1 amplification
ISF-T-013	FGFR2 fusions
ISF-T-014	FGFR2 amplification
ISF-T-015	FGFR3 fusions
ISF-T-016	FGFR3 amplification

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ISF-T-017	BCL2 fusions
ISF-T-018	BCL6 fusions
ISF-T-019	NRG1 fusions
ISF-T-020	MYC fusions
ISF-T-021	MYC amplification
ISF-T-022	1p/19q loss
ISF-T-023	CCNE1 amplification
ISF-T-024	MAF amplification

MAIN PRONOSTIC AND PREDICTIVE INMUNOHISTOCHEMISTRY (IHC) ASSAYS

CODE	SERVICE
ISI-T-001	ALK (D5F3)**
ISI-T-002	PDL1 (22C3)
ISI-T-003	Mismatch Repair (MMR) status (MLH1, MSH2, MSH6, PMS2)
ISI-T-004	MET (SP44)
ISI-T-005	BRG1 (SMARCA4) (E8V5B)
ISI-T-006	ERBB2 (4B5)
ISI-T-007	MTAP (E5SR1I)

EXTERNAL SERVICES

CODE	SERVICE
PO-E-001	Mandatory Market/Patient Price (Invitrocue - ONCOPDO ORGANOIDES SERVICE)
PO-E-002	Oncotype DX
PO-E-005	MLH1-MET (MLH1 Methylation)
PO-E-006	NGS-FUSION (study of >1300 fusions)
PO-E-012	Signatera Initial Test: Includes MRD detection in the baseline samples (tissue and blood ctDNA)
PO-E-014	TSO500 Panel: Analyzes 523 genes for DNA and 55 genes for RNA. Includes SNVs, indels, CNVs, TMB and MSI*
PO-E-015	Microsatellite Instability (MSI)

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MOLECULAR TESTING OF TUMOR LIQUID BIOPSY SAMPLES

CODE	Recommended studies by pathology
CS-BL-001	Lung tumors* Gene sequencing including EGFR, KRAS, BRAF (NGS Somatic QIAseq DNA Custom Panel 19 genes). nCounter RNA Fusion Solid Tumor Panel for Liquid Biopsies (6 genes) (including ALK, ROS, RET)
MS-BL-001	Colorectal tumors* Gene sequencing including KRAS, BRAF, NRAS (NGS QIAseq DNA Custom-19 genes)
MS-BL-004	Breast and gynecological tumors* Gene sequencing including PIK3CA, AKT1, ESR1 (NGS QIAseq DNA Custom Breast_ Gynecological Panel-11 genes)

MULTIGENIC MOLECULAR PANELS

CODE	Study mutations (SNVs/indels) and copy number variation (CNVs)
MS-BL-004	NGS Somatic QIAseq DNA custom Breast-Gynecological Panel (11 genes). Detection of somatic alterations in 9 genes with clinical relevance in breast, gynecological, and prostate cancer. Genes: AKT, BRCA1, BRCA2, CTNNB1, ERBB2, ESR1, PALB2, PIK3CA, POLE, PTEN, TP53
MS-BL-001*	NGS Somatic QIAseq DNA Custom Panel (19 genes). Detection of somatic alterations in 19 genes with clinical relevance in cancer. Genes: ALK, BRAF, CDK4, CDK6, EGFR, ERBB2, ERBB4, FGFR1, IDH1, IDH2, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, ROS1, STK11, TP53. (Includes METΔ14 splicing variant)
CODE	**Study of fusion genes and gene expression signatures**
MS-BL-003	nCounter RNA Fusion Solid Tumor Panel for Liquid Biopsies (6 genes). Fusions: ALK, NRG1, NTRK1, RET, ROS1. Splicing variant: METΔ14* Most frequent rearrangements

INDIVIDUAL GENE MUTATIONS (TaqMan® allelic discrimination qPCR assay)

CODE	SERVICE
ISM-BL-001	Single mutation detection (specify mutation)
ISM-BL-002	EGFR exon 19 del and EGFR exon 21 (L858R, L861Q) mutations
ISM-BL-003	EGFR exon 19 de, EGFR exon 21 (L858R, L861Q) and EGFR exon 20 T790M mutations
ISM-BL-004	EGFR exon 20 resistance mutation T790M
ISM-BL-005	EGFR exon 20 resistance mutation C797S
ISM-BL-006	KRAS hotspot mutations (codons 12, 13 and 61)
ISM-BL-007	BRAF hotspot mutations (V600E, V600K, V600R)
ISM-BL-008	PIK3CA hotspot mutations (codons 542, 545 and 1047)
ISM-BL-009	ESR1 hotspot mutations (codons 380, 536, 537, 538)

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EXTERNAL SERVICES

CODE	SERVICE
PO-E-013	Signatera Monitoring Test: Includes MRD monitoring in blood ctDNA

EXTERNAL SERVICES (ONCOHEMATOLOGY)

CODE	SERVICE
PO-E-007	Haematology OncokitDX (OncopanelDX)
PO-E-008	IGHV (Somatic hypermutations)

GERMLINE MOLECULAR TESTING ASSOCIATED WITH INHERITED PREDISPOSITION SYNDROM

MULTIGENIC MOLECULAR PANELS

CODE	SERVICE
MS-G-001	NGS QIAseq Inherited Cancer Panel (28 genes, germline). Detection of somatic alterations in 28 genes with clinical relevance in hereditary cancer and DNA repair pathways. The types of alterations detected include point mutations, small deletions/insertions, and copy number variations (CNVs). Genes: APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, CTNNB1, EPCAM, FANCA, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PIK3CA, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53

EXTERNAL SERVICES

CODE	SERVICE
PO-E-009	PMS2 gene: long-range (LR-PCR) sequencing of exons 11 to 15
PO-E-010	Pancancer (study of more than 150 genes associated with hereditary cancer)
PO-E-011	One Mutation (Analysis of a germline mutation in any gene (confirmation in family)

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MOLECULAR TESTING FOR OPHTHALMOLOGICAL DISEASES

MULTIGENIC MOLECULAR PANELS

CODE	Recommended analysis
OF-BL-001	nCounter Gene Expression Lacrima Dx (24 genes). Genes: B2M, CCL4, CD46, CFB, CTSS, GAPDH, HLA-A, HLA-DMB, HLA-DRB3, IFITM1, IRF7, JAK1, LGALS3, MUC1, MX1, MYD88, OAZ1, PSMB8, RARRES3, SERPING1, STAT1, STAT2, TAP1, TAPBP
OF-BL-002	nCounter Gene Expression Lacrima Extended Dx (96 genes). Genes: ABCf1, ADA, AHR, ALAS1, ARHGDIB, ATG5, B2M, BAX, BCAP31, BCL10, C14orf166, CASP3, CCL4, CCND3, CCRL1, CD14, CD19, CD46, CD59, CD83, CD99, CEACAM6, CFB, CFH, CFI, CTNNB1, CTSC, CTSS, CXCR2, DUSP4, EEF1G, FADD, FKBP5, G6PD, GAPDH, GUSB, HLA-A, HLA-C, HLA-DMB, HLA-DPA1, HLA-DQB1, HLA-DRA, HLA-DRB1, HLA-DRB3, HPRT1, IFI16, IFI35, IFITM1, IFNAR1, IFNG, IKZF2, IKBKG, IL13RA1, IL32, IL6ST, IL8, IRF5, IRF7, JAK1, LGALS3, LTF, MAPK1, MUC1, MX1, MYD88, NFKB1, NFKBIA, OAZ1, PIGR, PLA2G2A, PLAU, PML, POLR1B, POLR2A, PPIA, PSMB10, PSMB5, PSMB7, PSMB8, PTK2, PYCARD, RARRES3, RUNX1, S100A9, SDHA, SERPING1, SLC2A1, SMAD3, STAT1, STAT2, TAP1,TAPBP, TBP, TUBB

EXTERNAL SERVICES

CODE	SERVICE
PO-E-004	CentoVision Panel- 378 genes (Inherited Diseases)

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Genomic Sequencing Services for Research

We offer high-quality sequencing services for research projects, including Whole Exome Sequencing (WES), Whole Transcriptome Sequencing (WTS), and Whole Genome Sequencing (WGS). Optional bioinformatic analyses can be performed, and data is provided for download in standard formats.

Genomic Sequencing Services for Research
Whole Exome Sequencing

Service	DNA Type	Library Preparation	Sequencing	Output	Data Delivery	Bioinformatics (Optional)
WES Basic	HMW DNA	Human Core Exome Kit + RefSeq & Mitochondrial Panel	Illumina 2x100 bp	6 Gb per sample	FASTQ	Levels 2–5 optional
WES Premium	Low input / fragmented	Human Core Exome Kit + RefSeq & Mitochondrial Panel	Illumina 2x100 bp	12 Gb per sample	FASTQ	Levels 2–5 optional
WES Premium Deep	Low input / fragmented	Human Core Exome Kit + RefSeq & Mitochondrial Panel	Illumina 2x100 bp	24 Gb per sample	FASTQ	Levels 2–5 optional

Level	Description	Format
2	Read mapping	BAM, CSV (metrics)
3	SNV & small indel calling	VCF, CSV (metrics)
4	Variant annotation	JSON, TSV
5	Copy number variation (CNV) calling	VCF, GFF3, CSV (metrics)

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Genomic Sequencing Services for Research - Whole Transcriptome Sequencing

Service	Sample Number	Library Preparation	Sequencing	Output	Data Delivery	Bioinformatics (Optional)
WTS Classic	50 (2 batches of 25)	rRNA & globin mRNA depletion	Illumina 2x100 bp	30 M read pairs (~6 Gb) per sample	FASTQ	Levels 2–4 optional

Level	Description	Format
2	Read mapping	BAM
3	Read count normalization	TSV
4	Group comparison & differential expression analysis	TSV, PDF (heatmap, MA plot, volcano plot)

Genomic Sequencing Services for Research Whole Genome Sequencing

Service	DNA Type	Library Preparation	Sequencing	Output	Data Delivery	Bioinformatics (Optional)
WGS Large Classic (PCR-free)	HMW DNA	PCR-free library prep	Illumina 2x150 bp	90 Gb per sample	FASTQ	Levels 2–5 optional
HiFi WGS	HMW DNA	PCR-free library prep	**Long Reads**	60 Gb per sample	FASTQ	Levels 2–5 optional

Level	Description	Format
2	Read mapping & sex chromosome ploidy prediction	BAM, CSV
3	SNV & small indel calling, regions of homozygosity (ROH)	VCF, BED, CSV
4	SNV & indel annotation	JSON, TSV
5	CNV & structural variant (SV) calling, including translocations, inversions, large/medium indels	VCF, JSON, TSV, CSV

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