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ONCOLOGY

NOVEMBER 2026

SERVICES PORTFOLIO



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MOLECULAR TESTING OF TUMOR TISSUE SAMPLES

CODE	RECOMMENDED STUDY (ALL PATHOLOGIES)	TAT[1]
MS-T-009	**NGS TSO 500.** Detection of somatic alterations in 523 clinically relevant cancer genes using next-generation sequencing (NGS) panel. Includes analysis of single nucleotide variants (SNVs), insertions and deletions (indels), splicing variants, amplifications, gene fusions (55), homologous recombination deficiency (HRD; reported as Genomic Instability Score [GIS]), tumor mutational burden (TMB), and microsatellite instability (MSI). Genes included for SNVs, INDELs and CNVs (DNA): *ABL1, ABL2, ACVR1, ACVR1B, AKT1, AKT2, AKT3, ALK, ALOX12B, ANKRD11, ANKRD26, APC, AR, ARAF, ARFRP1, ARID1A, ARID1B, ARID2, ARID5B, ASXL1, ASXL2, ATM, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, AXL, B2M, BAP1, BARD1, BBC3, BCL10, BCL2, BCL2L1, BCL2L11, BCL2L2, BCL6, BCOR, BCORL1, BCR, BIRC3, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRIP1, BTG1, BTK, C11orf30, CALR, CARD11, CASP8, CBFB, CBL, CCND1, CCND2, CCND3, CCNE1, CD274, CD276, CD74, CD79A, CD79B, CDC73, CDH1, CDK12, CDK4, CDK6, CDK8, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CEBPA, CENPA, CHD2, CHD4, CHEK1, CHEK2, CIC, CREBBP, CRKL, CRLF2, CSF1R, CSF3R, CSNK1A1, CTCF, CTLA4, CTNNA1, CTNNB1, CUL3, CUX1, CXCR4, CYLD, DAXX, DCUN1D1, DDR2, DDX41, DHX15, DICER1, DIS3, DNAJB1, DNMT1, DNMT3A, DNMT3B, DOT1L, E2F3, EED, EGFL7, EGFR, EIF1AX, EIF4A2, EIF4E, EML4, EP300, EPCAM, EPHA3, EPHA5, EPHA7, EPHB1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ERRFI1, ESR1, ETS1, ETV1, ETV4, ETV5, ETV6, EWSR1, EZH2, FAM123B, FAM175A, FAM46C, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FAT1, FBXW7, FGF1, FGF10, FGF14, FGF19, FGF2, FGF23, FGF3, FGF4, FGF5, FGF6, FGF7, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLI1, FLT1, FLT3, FLT4, FOXA1, FOXL2, FOXO1, FOXP1, FRS2, FUBP1, FYN, GABRA6, GATA1, GATA2, GATA3, GATA4, GATA6, GEN1, GID4, GLI1, GNA11, GNA13, GNAQ, GNAS, GPR124, GPS2, GREM1, GRIN2A, GRM3, GSK3B, H3F3A, H3F3B, H3F3C, HGF, HIST1H1C, HIST1H2BD, HIST1H3A, HIST1H3B, HIST1H3C, HIST1H3D, HIST1H3E, HIST1H3F, HIST1H3G, HIST1H3H, HIST1H3I, HIST1H3J, HIST2H3A*, HIST2H3C*, HIST2H3D, HIST3H3, HLA-A*, HLA-B*, HLA-C*, HNF1A, HNRNPK, HOXB13, HRAS, HSD3B1, HSP90AA1, ICOSLG, ID3, IDH1, IDH2, IFNGR1, IGF1, IGF1R, IGF2, IKBKE, IKZF1, IL10, IL7R, INHA, INHBA, INPP4A, INPP4B, INSR, IRF2, IRF4, IRS1, IRS2, JAK1, JAK2, JAK3, JUN, KAT6A, KDM5A, KDM5C, KDM6A, KDR, KEAP1, KEL, KIF5B, KIT, KLF4, KLHL6, KMT2B*, KMT2C*, KMT2D*, KRAS, LAMP1, LATS1, LATS2, LMO1, LRP1B, LYN, LZTR1, MAGI2, MALT1, MAP2K1, MAP2K2, MAP2K4, MAP3K1, MAP3K13, MAP3K14, MAP3K4, MAPK1, MAPK3, MAX, MCL1, MDC1, MDM2, MDM4, MED12, MEF2B, MEN1, MET, MGA, MITF, MLH1, MLL, MLLT3, MPL, MRE11A, MSH2, MSH3, MSH6, MST1, MST1R, MTOR, MUTYH, MYB, MYC, MYCL1, MYCN, MYD88, MYOD1, NAB2, NBN, NCOA3, NCOR1, NEGR1, NF1, NF2, NFE2L2, NFKBIA, NKX2-1, NKX3-1, NOTCH1, NOTCH2, NOTCH3, NOTCH4, NPM1, NRAS, NRG1, NSD1, NTRK1, NTRK2, NTRK3, NUP93, NUTM1, PAK1, PAK3, PAK7, PALB2, PARK2, PARP1, PAX3, PAX5, PAX7, PAX8, PBRM1, PDCD1, PDCD1LG2, PDGFRA, PDGFRB, PDK1, PDPK1, PGR, PHF6, PHOX2B, PIK3C2B, PIK3C2G, PIK3C3, PIK3CA, PIK3CB, PIK3CD, PIK3CG, PIK3R1, PIK3R2, PIK3R3, PIM1, PLCG2, PLK2, PMAIP1, PMS1, PMS2, PNRC1, POLD1, POLE, PPARG, PPM1D, PPP2R1A, PPP2R2A, PPP6C, PRDM1, PREX2, PRKAR1A, PRKCI, PRKDC, PRSS8, PTCH1, PTEN, PTPN11, PTPRD, PTPRS, PTPRT, QKI, RAB35, RAC1, RAD21, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54L, RAF1, RANBP2, RARA, RASA1, RB1, RBM10, RECQL4, REL, RET, RFWD2, RHEB, RHOA, RICTOR, RIT1, RNF43, ROS1, RPS6KA4, RPS6KB1, RPS6KB2, RPTOR, RUNX1, RUNX1T1, RYBP, SDHA, SDHAF2, SDHB, SDHC, SDHD, SETBP1, SETD2, SF3B1, SH2B3, SH2D1A, SHQ1, SLIT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA4, SMARCB1, SMARCD1, SMC1A, SMC3, SMO, SNCAIP, SOCS1, SOX10, SOX17, SOX2, SOX9, SPEN, SPOP, SPTA1, SRC, SRSF2, STAG1, STAG2, STAT3, STAT4, STAT5A, STAT5B, STK11, STK40, SUFU, SUZ12, SYK, TAF1, TBX3, TCEB1, TCF3, TCF7L2, TERC, TERT, TET1, TET2, TFE3, TFRC, TGFBR1, TGFBR2, TMEM127, TMPRSS2, TNFAIP3, TNFRSF14, TOP1, TOP2A, TP53, TP63, TRAF2, TRAF7, TSC1, TSC2, TSHR, U2AF1, VEGFA, VHL, VTCN1, WISP3, WT1, XIAP, XPO1, XRCC2, YAP1, YES1, ZBTB2, ZBTB7A, ZFHX3, ZNF217, ZNF703, ZRSR2.* *Not covered for CNVs Genes included for Fusions (RNA): *ABL1, AKT3, ALK, AR, AXL, BCL2, BRAF, BRCA1, BRCA2, CDK4, CSF1R, EGFR, EML4, ERBB2, ERG, ESR1, ETS1, ETV1, ETV4, ETV5, EWSR1, FGFR1, FGFR2, FGFR3, FGFR4, FLI1, FLT1, FLT3, JAK2, KDR, KIF5B, KIT, MET, MLL, MLLT3, MSH2, MYC, NOTCH1, NOTCH2, NOTCH3, NRG1, NTRK1, NTRK2, NTRK3, PAX3, PAX7, PDGFRA, PDGFRB, PIK3CA, PPARG, RAF1, RET, ROS1, RPS6KB1, TMPRSS2.*	15

CODE	RECOMMENDED STUDIES BY PATHOLOGY	TAT[1]
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	10-12
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*)	

***See multigenic molecular panels.*

TISSUE SAMPLE SHIPMENT: All molecular assays in solid tumors require the shipment of Formalin Fixed Paraffin-Embedded (FFPE) blocks. Please contact us if the FFPE block is not available.

MULTIGENIC MOLECULAR PANELS

CODE	STUDY MUTATIONS (SNVS/INDELS) AND COPY NUMBER VARIATION (CNVS)	TAT[1]
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)	10-12
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 19 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**	**TAT[1]**
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*	8-10
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)	10-12

INDIVIDUAL GENE MUTATIONS [Targeted single-gene mutation analysis (qPCR)]

CODE	SERVICE	TAT[1]
ISM-T-002	*EGFR* exon 19 del and *EGFR* exon 21 (L858R, L861Q) mutations	5-7
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)	
ISM-T-010	*POLE* (exon 9: P286H/L/R/S, M295R, S297F; exon 11: F367S/V, D368Y; exon 13: V411L [G>C/G>T], L424I/V, P436R, M444K; exon 14: A456P, S459F) and *POLD1* (exon 12: S478N)	

ANATOMICAL PATHOLOGY DIAGNOSIS

CODE	SERVICE	TAT[1]
PP-T-003	Anatomical pathology diagnosis (H&E and 4 IHC analysis)	5-7
PP-T-004	Confirmation of anatomical pathology diagnosis (Previous AP report required)	2-3

TISSUE SAMPLE SHIPMENT: All molecular assays in solid tumors require the shipment of Formalin Fixed Paraffin-Embedded (FFPE) blocks. Please contact us if the FFPE block is not available.

MAIN FISH ASSAYS*

CODE	SERVICE	TAT[1]
ISF-T-001	*HER-2/neu* amplification**	5-7
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	
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	

**Ask for other IHC or FISH assays if required. **FDA approved.*

MAIN PRONOSTIC AND PREDICTIVE INMUNOHISTOCHEMISTRY (IHC) ASSAYS*

CODE	SERVICE	TAT[1]
ISI-T-001	ALK (D5F3)**	3-5
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)	
ISI-T-010	FOLR1 (FOLR1-2.1)	

**Ask for other IHC or FISH assays if required. **FDA approved.*

TISSUE SAMPLE SHIPMENT: All molecular assays in solid tumors require the shipment of Formalin Fixed Paraffin-Embedded (FFPE) blocks. Please contact us if the FFPE block is not available.

EXTERNAL SERVICES

CODE	SERVICE	TAT[1]
PO-E-002	Oncotype DX	15
PO-E-005	*MLH1*-MET (*MLH1* Methylation)	7-10
PO-E-006	NGS-FUSION (study of >1300 fusions)**	12-15
PO-E-012	**Signatera Initial Test:** Includes MRD detection in the baseline samples (tissue and blood ctDNA)	30
PO-E-015	Microsatellite Instability (MSI)	7

TISSUE SAMPLE SHIPMENT: All molecular assays in solid tumors require the shipment of Formalin Fixed Paraffin-Embedded (FFPE) blocks. Please contact us if the FFPE block is not available.

MOLECULAR TESTING OF TUMOR LIQUID BIOPSY SAMPLES

CODE	RECOMMENDED STUDIES BY PATHOLOGY	TAT[1]
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)*	10-12
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)	

**See details of the genes included in the multiplex panel section.*

MULTIGENIC MOLECULAR PANELS

CODE	STUDY MUTATIONS (SNVS/INDELS) AND COPY NUMBER VARIATION (CNVS)	TAT[1]
MS-BL-004	**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*	10-12
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**	**TAT[1]**
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	10-12

**MS-BL-001 could be replaced by the NGS Somatic QIAseq DNA Custom Extended Panel (30 genes) in specific cases where necessary (ask for prices)*

INDIVIDUAL GENE MUTATIONS [Targeted single-gene mutation analysis (qPCR)]

CODE	SERVICE	TAT[1]
ISM-BL-002	***EGFR*** **exon 19 del and *EGFR* exon 21 (L858R, L861Q) mutations**	5-7
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)**	

LIQUID BIOPSY SAMPLES SHIPMENT: For circulating liquid biopsy studies, two 10 mL Cell-Free DNA BCT (Streck) tubes are routinely required. When samples are shipped by express courier and received by the laboratory on the same day, three 10 mL EDTA tubes may be used as an alternative. Please contact us for assays on other types of biological fluids.

EXTERNAL SERVICES

CODE	SERVICE	TAT[1]
PO-E-013	**Signatera Monitoring Test:** Includes MRD monitoring in blood ctDNA	15
PO-E-017	**FoundationOne® Liquid Cdx.** It interrogates 324 genes, including 309 genes with complete exonic (coding) coverage and 15 genes with only select non-coding coverage.	10-15
PO-E-018	**FoundationOne® Heme.** It interrogates 406 genes with complete exonic (coding) coverage, including selected intronic coverage of 31 genes involved in rearrangements, and RNA sequencing of 265 commonly rearranged genes for fusion detection.	20-22
PO-E-019	**Tempus xF.** 105 gene liquid biopsy circulating tumor DNA (ctDNA) seq panel that detects oncogenic drivers and resistance mutations, and identifies single nucleotide variants (SNVs), insertions and deletions (INDELs), copy number gains (CNGs) and gene rearrangements, all of which help inform precision medicine decisions.	17
PO-E-020	**Tempus xF+.** 523 gene liquid biopsy panel, covering SNVs, INDELs, CNGs and gene rearrangements. Additionally, xF+ identifies variants that are potentially associated with clonal hematopoiesis of indeterminate potential (CH).	17

EXTERNAL SERVICES (ONCOHEMATOLOGY)

CODE	SERVICE	TAT[1]
PO-E-007	**Haematology OncokitDX (OncopanelDX).** Sequencing of whole exonic regions of the following genes: *ARID5B, ASXL1, ASXL2, ATRX, BCOR, BCORL1, BLNK, BRAF, CALR, CBL, CDKN2A, CDKN2B, CEBPA, CHIC2, CREBBP, CRLF2, CSF3R, CSNK1A1, CUX1, DDX3X, DDX41, DNMT3A, EP300, ETNK1, ETV6, EZH2, FBXW7, FLT3, GATA1, GATA2 , GATA3, HAVCR2, IDH1, IDH2, IKZF1, IL7R, JAK1, JAK2, JAK3, KIT, KMT2A, KRAS, MPL, NF1, NFE2, NOTCH1, NPM1, NR3C1, NRAS, PAX5, PHF6, PIGA, PPM1D, PTEN, PTK2B, PTPN11, RAD21, RB1, RUNX1, SETBP1, SF3B1, SH2B3, SAMD9L, SMC1A, SMC3, SRP72, SRSF2, STAG1, STAG2, STAT5B, TERT, TET2, TPMT, TP53, TYK2, U2AF1, WT1, ZRSR2. Sequencing of non-coding regions and rearrangements: ABL1, ABL2, BCR, CBFA2T3, CBFB, CSF1R, EPOR, ETV6, FGFR1, FUS, JAK2, KMT2A , MEF2D , MNX1, NPM1, NUP214, NUP98, PDGFRA, PDGFRB, RARA, RBM15, RUNX1, TAL1, TCF3*	12
PO-E-008	**IGHV (Somatic hypermutations)**	10-12

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GERMLINE MOLECULAR TESTING ASSOCIATED WITH INHERITED PREDISPOSITION SYNDROM

MULTIGENIC MOLECULAR PANELS

CODE	SERVICE	TAT[1]
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*	10-12

EXTERNAL SERVICES

CODE	SERVICE	TAT[1]
PO-E-009	***PMS2* gene:** long-range (LR-PCR) sequencing of exons 11 to 15	25
PO-E-011	**One Mutation** (Analysis of a germline mutation in any gene (confirmation in family)	
PO-E-016	**Hereditary Plus OncoKitDx (101 genes)** *Target genes (full sequencing and CNV analysis): *ABRAXAS1 (FAM175A), ACD, AIP, AKT1, ALK, APC (incl. 5' UTR), ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CHEK2, CTNNA1, CYLD, DICER1, EPCAM (incl. 3' UTR), FANCC, FANCG, FANCM, FH, FLCN, GALNT12, GALNT14, GDNF, GEN1, GREM1, HABP2, HOXB13, KIF1B, KLLN, LZTR1, MAX, MC1R, MEN1, MET, MITF, MLH1, MLH3, MRE11A, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RB1, RECQL4, RET, RINT1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPRED1, STK11, SUFU, TERF2IP, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WT1* y *XRCC2.* Additional CNV regions: *EPCAM (3' UTR)* y *MSH2 (5' UTR)* 50 hotspots in the following genes: *APC, ATM, ATR, BRCA1, BRCA2, CHEK2, FH, LZTR1, MET, MLH1, MSH2, NF1, PMS2, PTEN, RB1, RET, SDHB, STK11* y *TERT*	20-25

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MOLECULAR TESTING FOR OPHTHALMOLOGICAL DISEASES

MULTIGENIC MOLECULAR PANELS

CODE	RECOMMENDED ANALYSIS	TAT[1]
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*	10-12
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*	10-12

EXTERNAL SERVICES

CODE	SERVICE	TAT[1]
PO-E-004	**CentoVision Panel- 378 genes (Inherited Diseases):** Achromatopsia, Albinism, Bardet-Biedl syndrome, Cataract, Cone-rod and cone dystrophy, Flecked retina, Glaucoma, Hermansky-Pudlak syndrome, Leber congenital amaurosis, Meckel syndrome, Microphthalmia/anophthalmia/ coloboma spectrum, Oculomotor apraxia, Optic atrophy, Progressive external ophthalmoplegia, Retinitis pigmentosa, autosomal dominant, Retinitis pigmentosa, autosomal recessive, Stargardt disease, Stickler síndrome, Usher syndrome, Vitreoretinopathy, Wagner syndrome.	25

Prices and service offerings in this portfolio will be reviewed annually.
NOTE: If you need more specific information about any of the tests, please contact lab@panoncology.com

[1]Turnaround time (TAT): the time required to process and issue the report. TAT is measured in business days from the sample's receipt in our Lab.

PANGAEA LAB

OPHTHALMOLOGICAL DISEASE SAMPLES SHIPMENT: One EDTA tube of blood is routinely required for most liquid biopsy assays. For tear samples, please contact the laboratory prior to shipment at lab@panoncology.com.