

Reference Standard Pipeline

Products	Gene information	Allelic Frequencies	Format	관련제품
Quantitative Multiplex NGS gene panel validation을 위한 oncology reference standard	BRAF, KIT, EGFR, KRAS, NRAS, PI3KCA	1-24.5 %	gDNA, FFPE, fcDNA	HD701, HD200, HD798, HD799, HD803
Structural Multiplex Structural variant 확인을 위한 다양한 variant type 보유	AKT1, BRCA2, EGFR, FBXW7, FLT3, GNA11, KRAS, MET, MYC, NOTCH1, PIK3CA, RET, ROS1	4 % - 6 %	gDNA, cfDNA (in buffer)	HD753, HD786
Cell free DNA Fragmented reference standard (size 160-170 bp)	AKT1, BRCA2, EGFR, FBXW7, FLT3, GNA11, KRAS, MET, MYC, NOTCH1, NRAS, PIK3CA, RET, ROS1 중 일부	0 %, 0.1 %, 1 %, 5 % or 4-6 %	cfDNA in buffer or synthet- ic plasma	HD780, HD786, HD816, HD817, HD825
OncoSpan NGS 및 ddPCR로 검증된 reference standard	NGS로 검증된 386 variants (152 oncology genes) ddPCR로 검증된 25 variants (16 oncology genes)	1 % - 92 % for ddPCR- verified variants	gDNA, FFE, cfDNA	HD827, HD832, HD833
Tru-Q LOD 확인을 위해 원하는 Allelic Frequency 조제 가능	ABL1, ALK, BRAF, EGFR, FGFR2, FLT3, GNA11, GNAQ IDH1, IDH2, JAK2, KIT, KRAS, MEK, MET, NOTCH1, NRAS, PDGFRA, PIK3CA 중 일부	0-5 %	gDNA	HD728, HD729, HD730, HD731, HD734 HD752: 40 wild type loci 4 endogenous variants)
Single Gene Singleplex One variant in one gene	ABL1, AKT1, ALK, EML4-ALK, BRAF, EGFR, FGFR2, FTL3, GNA11, GNAQ, GNAS, IDH1, IDH2, JAK2, KIT, KRAS, MEK1, MET, NOTCH1, NRAS, PDGFRA, PI3KCA, PTEN중 한가지	1 %, 5 %, 20 %, 33 %, 50 %, 80 %, 100 %	FFPE, DNA	홈페이지 참조
RNA fusion RNA fusion을 연구를 위한 reference standard			FFPE	HD783: Negative for EML4-ALK, CCDC6-RET, SLC34A2-ROS1, TPM3-NTRK1, ETV6-NTRK3 HD784: ALK-RET ROS1 Fusion
Single-gene multiplex Wild type을 이용하여 원하는 Allelic Freauency 조제 가능	EGFR, KRAS중 한가지	0-12.5 %	gDNA, FFPE	HD850, HD300, HD802, HD301 HD709: EGFR wild type DNA HD710: KRAS wild type DNA
Myeloid DNA 골수암 관련 유전자 reference standard	ABL1, ASXL1, BCOR, CBL, DNMT3A, EZH2, FLT3, GATA1, GATA2, IDH1, IDH2, JAK2, KRAS NPM1, NRAS, RUNX1, SF3B1, TET2, TP53	5-70 %	gDNA	HD829
BRCA BRCA test용 reference standard	BRCA1, BRCA2, BRIP1, BRAD1, NBN	Germline: 50, 100 %, somatic: 7.5 – 100 %	gDNA, FFPE	HD793, HD794, HD795, HD810
MSS & MSI MSI, MSS 관련 20 종류의 biomarker 포함	Biomarkers: BAT-26, BAT-25, MONO-27, NR-21, NR-24, BTBD7, DDO1, ACVR2A, SEC31A, MRE11, SULF2, RYR3, BAT-40, TGFβ-11, D2S123, D18S58, D5S346, CAT-25, NR-22, NR-27		FFPE	HD830, HD831
Genome in a Bottle Cell Lines (GIAB) Whole genome sequencing을 위한 reference standard			FFPE	GM24149, GM24143, GM24385, GM24631