

Molecular Reference Standards

Horizon Discovery사는 정확한 유전자 편집 방법을 통해 생산한 Isogenic한 mutant cell line을 이용하여 제작한 다양한 표준 물질*을 제공합니다.

다양한 형태로 제공

gDNA, FFPE, RNA, cfDNA,
formalin-compromised DNA(fcDNA)

Cell line으로부터 제작하여
일정한 Genomic Complexity 유지

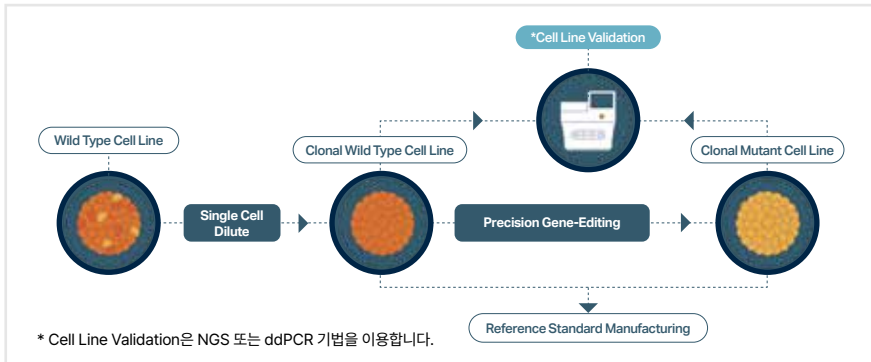
자제적인 cell line engineering을 통해
원하는 유전적 변이 선택 가능

국제표준화기구에서 인증받은 개발 및 제조 과정
ISO 13485:2016, ISO 9001:2015

* 표준 물질 (Reference Standard)이란?

특징이 명확하고 표준화되어 있으며 검증이 완료된 물질로, 실험 과정 또는 결과 정확도 판단 기준으로 활용됩니다.

Reference Standards 제작 과정



Reference Standard 제품 구성

Genomic DNA (gDNA)

- 다양한 assay에서 detection 기준 확립 시 용이한 형태
- Copy number variation 및 allelic frequency 확인 시 활용 가능

Cell-free DNA (cfDNA)

- 액체 생검(liquid biopsy) 과정에서 사용되는 DNA 형태
- 합성 플라즈마 또는 버퍼에 담긴 상태로 제공

Formalin-fixed reference standards (FFPE)

- FFPE 샘플의 가변성 확인을 위한 control 물질로 활용
- RNA fusions된 FFPE 제공 (Fusion Negative Control과 함께 이용 가능)

Formalin-compromised DNA (fcDNA)

- 포르말린에 의해 손상된 DNA를 취급하는 assay 검증 시 활용
- 손상 정도에 따른 3가지 타입 제공 (Mild / Moderate / Severe controls)

Application별 Reference Standards

NGS Validation

표준 물질을 활용한 패널 개발 최적화 및 정기적 분석 성능 모니터링은 분석 결과의 신뢰도를 높일 수 있습니다.

NGS Assay	Somatic/Structural Mutation 검출을 위한 Reference Standards			
Variant	Genes	Format	PN	Product
Single-gene Multiplex	BRCA	gDNA	HD795	BRCA Somatic Multiplex I (gDNA)
		FFPE	HD810	BRCA Somatic Multiplex I (FFPE)
	EGFR	FFPE	HD300	EGFR Gene-Specific Multiplex Reference Standard (FFPE) 5% AF
			HD850	EGFR Gene-Specific Multiplex Reference Standard (FFPE) 1% AF
		gDNA	HD802	EGFR Gene-Specific Multiplex Reference Standard (gDNA)
		KRAS	FFPE	HD301
Quantitative Multiplex	Multigenes	FFPE	HD200	Quantitative Multiplex Reference Standard (FFPE)
		gDNA	HD701	Quantitative Multiplex Reference Standard (gDNA)
Structural Multiplex		FFPE	HD789	Structural Multiplex Reference Standard (FFPE)
		gDNA	HD753	Structural Multiplex Reference Standard (gDNA)
		cfDNA	HD786	Structural Multiplex cfDNA Reference Standard

NGS Assay	Formalin-compromised DNA 손상 정도에 따른 Reference Standards			
Variant	Genes	Format	PN	Product
Quantitative Multiplex	Multigenes	fcDNA	HD798	Quantitative Multiplex Reference Standard fcDNA (mild)
			HD799	Quantitative Multiplex Reference Standard fcDNA (moderate)
			HD803	Quantitative Multiplex Reference Standard fcDNA (severe)

NGS Assay	Germline mutation 검출을 위한 Reference Standards			
Genes	Format	PN	Product	
BRCA	gDNA	HD793	BRCA Germline I (gDNA)	
		HD794	BRCA Germline II (gDNA)	

Tru-Q	LOD 확인을 위해 원하는 Allelic Frequency 조제 가능한 Reference Standards			
Genes	Format	PN	Product	
Multigenes	gDNA	HD728	Tru-Q 1 (5% Tier) Reference Standard	
		HD729	Tru-Q 2 (5% Tier) Reference Standard	
		HD730	Tru-Q 3 (5% Tier) Reference Standard	
		HD731	Tru-Q 4 (5% Tier) Reference Standard	
		HD734	Tru-Q 7 (1.3% Tier) Reference Standard	
		HD752	Tru-Q 0 (100% Wild type) Reference Standard	

GIAB	Whole Genome Sequencing 개발 및 검증을 위한 Reference Standards			
Genes	Format	PN	Product	
Multigenes	FFPE	GM24143	Genome in a Bottle Ashkenazim PGP Mother Reference Standard	
		GM24149	Genome in a Bottle Ashkenazim PGP Father Reference Standard	
		GM24385	Genome in a Bottle Ashkenazim PGP Son Reference Standard	
		GM24631	Genome in a Bottle Asian PGP Son Reference Standard	

Molecular Reference Standards

Application별 Reference Standards

Multiple Variant

액체 생검, 암 진단, MRD 검사 등에 필요한 다양한 종류의 표준 물질을 하나의 제품으로 제공합니다.

Gene Fusion			
RNA fusion 연구를 위한 Reference Standards			
Genes	Format	PN	Product
Multigenes	FFPE	HD784	ALK RET ROS RNA fusion
		HD834	Pan-Cancer 6-Fusion Panel
		HD835	Pan-Cancer 6-Fusion Panel (Negative Control)

cfDNA				
Cell-free DNA assay 성능 평가를 위한 Reference Standards				
Genes	Format	PN	Product	Available Assay
Multigenes	cfDNA	HD780	Multiplex I cfDNA Reference Standard Set	cfDNA NGS, qPCR assays
		HD917	Multiplex I cfDNA in Synthetic Matrix II	cfDNA recovery를 위한 표준 물질
Aneuploidy	cfDNA	HD863	Trisomy 21 Female-Matched Reference Standard	NIPT tests
		HD862	Trisomy 18 Male-Matched Reference Standard	
		HD861	Trisomy 13 Female-Matched Reference Standard	
		HD860	Euploid Male-Matched Reference Standard	

Oncology					
Cancer 관련 다양한 assay 성능 평가를 위한 Reference Standards					
Genes	Format	PN	Product	Available Assay	
Large Oncology & Exome	gDNA	HD827	OncoSpan gDNA	NGS (386 variants, 152 oncology genes) ddPCR (25 variants, 16 oncology genes)	
	FFPE	HD832	OncoSpan FFPE		
	cfDNA	HD833	Oncospan cfDNA		
Myeloid	gDNA	HD829	Myeloid DNA Reference Standard	Myeloid sequencing assays	
	cfDNA	HD838	Myeloid Panel	Liquid biopsy-based MRD assays	
		HD839	Myeloid Panel Negative Control	Liquid biopsy-based MRD assays	
Prostate Cancer	cfDNA	HD836	Prostate Cancer Panel	Prostate cancer sequencing assays	
		HD837	Prostate Cancer Panel (Negative Control)	Prostate cancer sequencing assays	
Colorectal Cancer	cfDNA	HD842	Colorectal Cancer ctDNA panel	MRD assay	
	cfDNA	HD843	Colorectal Cancer ctDNA panel negative control	MRD assay	
Non-Small Cell Lung Cancer	cfDNA	HD844	NSCLC ctDNA multiplex panel	MRD assay	
		HD845	NSCLC ctDNA multiplex panel negative control	MRD assay	
Lymphoid Cancer	FFPE	HD849	Lymphoid Cancer Panel FFPE	Molecular assay (Lymphoma cancer)	
		gDNA	HD840	Lymphoid Panel	Diagnostic assay (Lymphoma cancer)
			HD841	Lymphoid Panel Negative Control	Diagnostic assay (Lymphoma cancer)
Phosphate and tensin homolog	FFPE	HD864	PTEN Loss FFPE	Companion diagnostic (CDx) tests	

Application별 Reference Standards

Single Variant

주요 암 관련 유전자에 대한 0 – 100% 내 특정 대립 유전자 빈도의 물질로 제공합니다.
(BRAF, EGFR, FLT3, KRAS, PI3KCA, JAK2, NRAS, IDH1/2, EML4, AKT1)

Oncology		BRAF, EGFR, FLT3 유전자에 대한 Reference Standards			
Variant	Genes	Format	PN	Product	
Single-gene Singleplex	BRAF	FFPE	HD123	BRAF V600K Reference Standard, 5%	
			HD232	BRAF V600E Reference Standard, 20%	
			HD266	BRAF wild type FFPE, 100%	
			HD268	BRAF V600K Reference Standard, 50%	
			HD273	BRAF V600E Reference Standard, 1%	
			HD274	BRAF V600K Reference Standard, 1%	
			HD275	BRAF V600R Reference Standard, 50%	
		HD598	BRAF V600E FFPE Reference Standard, 50%		
		HD705	BRAF V600E Reference Standard, 5%		
		gDNA	HD238	BRAF V600E gDNA Reference Standard, 50%	
			HD249	BRAF wild type gDNA, 100%	
			HD125	EGFR T790M Reference Standard, 5%	
		EGFR	FFPE	HD126	EGFR T790M Reference Standard, 20%
				HD127	EGFR T790M FFPE Reference Standard, 50%
	HD129			EGFR L858R Reference Standard, 5%	
	HD130			EGFR L858R Reference Standard, 20%	
	HD131			EGFR L858R Reference Standard, 50%	
	HD132			EGFR L861Q Reference Standard, 5%	
	HD133			EGFR L861Q Reference Standard, 50%	
	HD141		EGFR Wild Type Reference Standard		
	HD230		EGFR G719S Reference Standard, 50%		
	HD308		EGFR E746-A750 FFPE, 50%		
	gDNA		HD653	EGFR E746-A750 Reference Standard, 5%	
			HD706	EGFR V769_D770insASV Reference Standard, 50%	
			HD707	EGFR S7681 FFPE, 50%	
		HD714	EGFR G719S Reference Standard, 5%		
		HD747	EGFR G719A Reference Standard, 50%		
HD251		EGFR E746-A750 Reference Standard, 50%			
cfDNA	HD254	EGFR L858R gDNA Reference Standard, 50%			
	HD258	EGFR T790M gDNA Reference Standard, 50%			
	HD260	EGFR A767_V769dup Reference Standard, 50%			
	HD261	EGFR S7681 gDNA Reference Standard, 50%			
	HD262	EGFR G719A Reference Standard, 50%			
FLT3	gDNA	HD709	EGFR Wild Type Reference Standard		
		HD825	EGFR Multiplex cfDNA Reference Standard Set		
		HD668	FLT3 D835Y Reference Standard, 50%		
			HD669	FLT3 Wild Type Reference Standard	

Molecular Reference Standards

Single Variant

Oncology	KRAS, PI3KCA 유전자에 대한 Reference Standards			
Variant	Genes	Format	PN	Product
Single-gene Singleplex	KRAS	FFPE	HD113	KRAS G13D Reference Standard, 5%
			HD117	KRAS G12A Reference Standard, 5%
			HD119	KRAS G12D Reference Standard, 5%
			HD135	KRAS Wild Type Reference Standard
			HD140	KRAS Q61L Reference Standard, 50%
			HD204	KRAS G12D Reference Standard, 50%
			HD207	KRAS G12R Reference Standard, 50%
			HD210	KRAS G12S Reference Standard, 50%
			HD213	KRAS G12V Reference Standard, 50%
			HD216	KRAS G13D Reference Standard, 50%
			HD255	KRAS G12A Reference Standard, 50%
			HD256	KRAS G12C Reference Standard, 50%
			HD715	KRAS A59T Reference Standard, 50%
			HD94	KRAS Q61H Reference Standard, 50%
			HD265	KRAS G12A Reference Standard, 50%
	HD269	KRAS G12C Reference Standard, 50%		
	gDNA	HD272	KRAS G12D Reference Standard, 50%	
		HD287	KRAS G12R Reference Standard, 50%	
		HD288	KRAS G12S Reference Standard, 50%	
		HD289	KRAS G12V Reference Standard, 50%	
		HD290	KRAS G13D Reference Standard, 50%	
		HD710	KRAS Wild Type Reference Standard	
	PI3KCA	FFPE	HD112	PIK3CA E545K Reference Standard, 50%
			HD121	PIK3CA E542K Reference Standard, 50%
			HD320	PIK3CA Wild Type Reference Standard
		HD599	PIK3CA H1047R Reference Standard, 50%	
		gDNA	HD688	PIK3CA E542K Reference Standard, 50%
			HD689	PIK3CA E545K Reference Standard, 50%
			HD690	PIK3CA H1047R Genomic Reference Standard, 50%
			HD691	PIK3CA Wild Type Reference Standard

Oncology	KRAS, PI3KCA 유전자에 대한 Reference Standards			
Variant	Genes	Format	PN	Product
Single-gene Singleplex	JAK2	gDNA	HD649	JAK2 V617F Reference Standard, 50%
			HD652	JAK2 Wild Type Reference Standard
			HD286	NRAS Q61R Reference Standard, 50%
	NRAS	FFPE	HD322	NRAS Wild Type Reference Standard
			HD745	NRAS G12D Reference Standard, 50%
			gDNA	HD648
	IDH1	gDNA	HD677	IDH1 R132H Reference Standard, 50%
			HD678	IDH1 Wild Type Reference Standard
	IDH2	gDNA	HD680	IDH2 R172K Reference Standard, 50%
			HD681	IDH2 Wild Type Reference Standard
	EML4	gDNA	HD664	EML4-ALK Reference Standard, 50%
	AKT1	FFPE	HD172	AKT1 Wild Type Reference Standard