

Structural Multiplex Reference Standards

Our Structural Multiplex Reference Standards cover a wide range of mutations in a defined genomic context. They are designed to challenge your molecular and bioinformatics protocols by providing validated CNVs/amplifications, translocations and large insertions/deletions, as well as SNPs/SNVs in high GC-rich regions.

Features

- Cell line-derived controls for the closest representation of your patient sample
- Available in gDNA, cfDNA and FFPE formats
- ddPCR /Sanger validation for 22 variants with defined allelic frequencies
- Includes RET and ROS1 fusion variants, MYC-N and MET focal amplifications and a BRCA2 variant

Applications

- Evaluate workflow integrity from pre-analytical DNA extraction to post-analytical bioinformatics with our FFPE product
- Optimize and validate the sensitivity of your workflow and the efficacy of your platform
- Optimize and validate new cancer panels and routinely monitor the performance of your assay

Genes and Variants

- 8 genes
- 9 variants
- 3 SNPs (SNVs), 2 fusions, 2 CNVs, 2 INDELS

Verified Mutations

Gene	Amino Acid Change	Chromosome	Genomic Position	Reference	Alternative	Variant Type	COSMIC ID	Expected AF (%)
AKT1	E17K	14	105246551	C	T	SNV High GC	COSM	5.0
EGFR	V769_D770insASV	7	55249010	-	GCCAGCGTG	Long Insertion	COSM	5.6
EGFR	ΔE746 - A750	7	55242465	-	AA	Long Deletion	COSM	5.3
GNA11	Q209L	19	3118942	A	T	SNV High GC	COSM	5.6
MET	amplification	7	chr.7	-	-	CNV	COSM	4.5 copies
MYCN	amplification	2	chr.2	-	-	CNV	COSM	9.5 copies
PIK3CA	E545K	3	178936091	G	A	SNV Low GC	COSM	5.6
RET	CCDC6/RET fusion	10		-	-	Fusion	COSM	5.0
ROS1	SLC34A2/ROS1 fusion	4:6		-	-	Fusion	N/A	5.6

Presence Confirmed in Parental Cell Line

Gene	Amino Acid Change	Chromosome	Genomic Position	Reference	Alternative	Variant Type	COSMIC ID	Expected AF (%)
BRAF	V600E	7	140453136	T	A	SNV	COSM	18.2
BRCA2	A1689fs	13	32913558	CA	C	Short Deletion	COSM	5.6
EGFR	G719S	7	55241707	G	A	SNV	COSM	5.3
FBXW7	G667fs	4	153244155	TC	T	Short Deletion	COSM	5.6
FLT3	S985fs	13	28578214	GGA	G	Short Deletion	COSM	5.6
KRAS	G13D	12	25398281	C	T	SNV Low GC	COSM	5.6
MET	V237fs	7	116339847	GT	G	Short Deletion	COSM	2.5
NOTCH1	P668S	9	139409754	G	A	SNV High GC	COSM	5.0
PIK3CA	H1047R	3	178952085	A	G	SNV	N/A	16.7

Product Specification

	FFPE	gDNA	cfDNA
Catalog Number	HD789	HD753	HD786
Format	FFPE	gDNA	cfDNA
Quantity	1 FFPE curl	1 μg	350ng
Concentration	N/A	50ng/μl	20ng/μl
Quality Certificate	ISO 9001, 13485	ISO 9001, 13485	ISO 9001

If you have any questions, contact

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