Structural Multiplex Reference Standards

Overview

Structural variants including gene translocations, fusions, copy number variants (CNVs) and INDELs are commonly found in tumors, and can serve as clinical biomarkers to identify cancer, to monitor disease progression and to guide cancer treatment.

However, the detection of structural variants presents several challenges and bioinformatics pipelines are continuously evolving to improve confidence in structural variant detection.

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 single-nucleotide polymorphisms (SNPs/SNVs) in high GC-rich regions.

Features

Key Benefits

- Cell line-derived controls for the closest representation of your patient sample
- Available in gDNA, cfDNA and FFPE formats
- ddPCR validation for 9 variants with defined allelic frequencies and copy number for the CNVs
- 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 Structural Multiplex
- Optimize and validate the sensitivity of your workflow for structural variant detection and the efficacy of your platform under GC/AT-rich regions
- 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

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Verified Mutations

Variant Type	Chromosome Number	Gene	Variant
SNV High GC	chr. 19	GNA11	Q209L
SNV High GC	chr. 14	AKT1	E17K
SNV Low GC	chr. 3	PIK3CA	E545K
Long Insertion	chr. 7	EGFR	V769_D770insASV
Long Deletion	chr. 7	EGFR	ΔE746-A750
Fusion	chr. 4: chr. 6	ROS1	SLC34A2/ROS1 fusion
Fusion	chr. 10	RET	CCDC6/RET fusion
CNV	chr. 7	MET	Amplification
CNV	chr. 2	MYC-N	Amplification

Presence Confirmed in Parental Cell Line

Variant Type	Chromosome Number	Gene	Variant
SNV Low GC	chr. 12	KRAS	G13D
SNV High GC	chr. 9	NOTCH1	P668S
Short Deletion	chr. 7	MET	V237fs
Short Deletion	chr. 13	FLT3	S985fs
Short Deletion	chr. 13	BRCA2	p.k1691fs*15
Short Deletion	chr. 4	FBXW7	G667fs
SNV	chr. 7	EGFR	G719S
SNV	chr. 7	BRAF	V600E
SNV	chr. 3	PIK3A	H1047R

Product Specification

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