PGDx

PlasmaSELECT™ 64 + MSI

The only pan-cancer assay for tumor profiling that can evaluate microsatellite status in plasma

PlasmaSELECT 64 TEST

- Identifies clinically actionable and functionally important sequence mutations and structural alterations across multiple cancer types without the need for invasive biopsies
- Reports microsatellite instability status (MSI) to assess potential response to checkpoint inhibitor therapies[1,2,3]

Microsatellite Instability Status

- Identifies sequence mutations (single base substitutions, indels), amplifications, and rearrangements with unparalleled accuracy
- How PGDx does it:
  - Proprietary DNA extraction and preparation methods that accommodate low-abundance cell-free DNA samples
  - Proprietary hybrid-capture processing in combination with high-coverage, next-generation sequencing
  - Proprietary VariantDx™ computational algorithms enable discrimination of sequencing artifacts and errors from bona-fide mutations
- Comprehensive clinical annotation of all reported alterations, including, FDA-approved therapies, clinical trials, and published literature

References

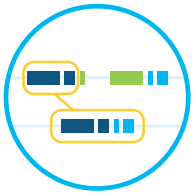
1. Le et al., 2015. N. Engl. J. Med. 2. Rizvi et al., 2015. Science 3. Snyder et al., 2014. N. Engl. J. Med.
4. Jones et al., 2015. Sci. Transl. Med.

PlasmaSELECT 64 IDENTIFIES:

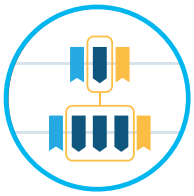
Sequence Mutations

Single Base Substitutions

Insertions and Deletions

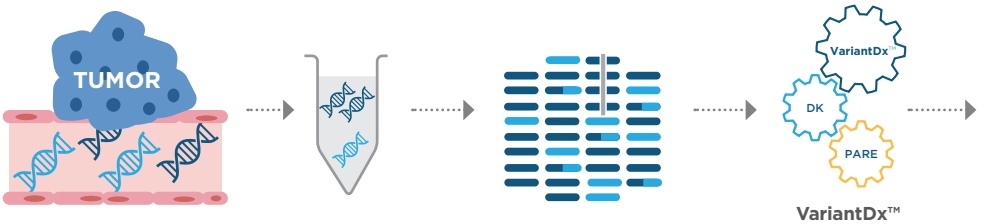


Genomic Rearrangements



Amplifications

SCHEMATIC FOR DETECTING GENETIC ALTERATIONS FROM PLASMA. The methods use next-generation paired-end sequencing of cell-free DNA isolated from plasma to identify genetic alterations characteristic of tumor DNA. Such alterations include rearrangements, sequence mutations, and amplifications. (from Leary et al, Sci Transl Med. 2012)

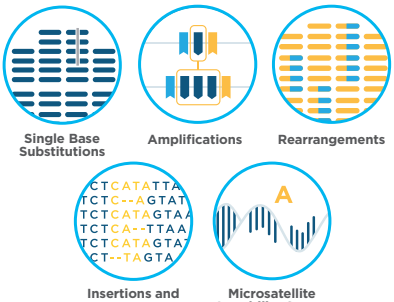


Tumor DNA entering circulation

Cell-free DNA isolated from plasma

Paired-end massively parallel sequencing

VariantDx™
Digital Karyotyping (DK)
Rearrangement analysis (PARE)



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BENEFITS OF USING PLASMA FOR CANCER MUTATION ANALYSIS

A non-invasive, liquid biopsy assay will be helpful for cancer patients when:

- Biopsy is not available, insufficient or exhausted before molecular testing can be performed
- Tissue-based molecular testing failed due to poor-quality DNA from tissue biopsy
- Suspicion that the tumor may have acquired mutations as result of treatment since the original biopsy was taken
- Desire to detect genetic alterations in multiple lesions simultaneously
- Desire to avoid the risks and costs associated with re-biopsy

GENES EVALUATED IN PlasmaSELECT 64

*Full coding and specific exon analyses in 58 well-characterized cancer genes, as well as †amplification analyses for 19 genes

AKT1	CCND2†	EGFR*†	GNAS	MET†	NTRK3	PTEN	TSC2
ALK*†	CCND3†	ERBB2*†	HRAS	MTOR	PALB2	RB1	VHL
AR*†	CD274*†	ESR1	IDH1	MYC†	PIK3CA†	RET*	
ATM	CDK4*†	EZH2	IDH2	MYCN†	PIK3CB	RNF43	
BRAF*	CDK6*†	FGFR1†	JAK2	NPM1	PIK3R1	ROS1†	
BRCA1	CDKN2A	FGFR2†	KIT*†	NRAS	POLD1	TERT	
BRCA2	CTNNB1	FGFR3†	KRAS*	NTRK1	POLE	TP53*	
CCND1†	DNMT3A	FLT3	MAP2K1	NTRK2	PTCH1	TSC1	

Rearrangement analyses for selected regions of 17 well-characterized cancer genes

ALK	EGFR	FGFR2	NTRK1	PDGFRB	RET
BCR	ETV6	FGFR3	NTRK2	RAF1	ROS1
BRAF	FGFR1	MYC	PDGFRA	RARA	

Microsatellite analysis for 5 well-characterized mononucleotide sequences

BAT-25	BAT-26	NR-21	NR24	MONO-27

PlasmaSELECT 64 ASSAY PERFORMANCE

Performance Specification	Mutant Allele Fraction	Sensitivity	Specificity
Sequence Mutations (SBS/Indel)	≥0.50%	99.4%	>99.999%*
Rearrangements	≥0.50%	94.4%	>99%
Microsatellite Instability Status (MSI)	≥2.0%	>99%	>99%
Amplifications (≥4-fold)	≥20%	97.2%	>99%
Amplifications (≥4-fold)	<20%	varies depending on level of amplification and tumor content	>99%

*Per-base specificity provided for sequence mutation analyses [99,359 bases evaluated]

PlasmaSELECT 64 SEQUENCING KEY METRICS

Sequencing Method	Illumina next-generation sequencing
Bioinformatics	Patented PARE, Digital Karyotyping and VariantDx
Turnaround Time	2-3 weeks
Sample Requirements	Whole blood or plasma
Plasma Sample Input Required	Two 10 ml tubes of peripheral whole blood; 6-10 ml plasma collected according to PGDx sample preparation instructions (>1 -2 ml for RU0)

PlasmaSELECT 64 SEQUENCING DELIVERABLES & ANALYSES

Regions Analyzed	64 genes
Sample Prep and NGS Sequencing	
Sequence Mutation Analysis	
Rearrangement Analysis	
Amplification Analysis	
Microsatellite Instability Status	
Clinical Annotation	

Related References
Diaz et al. Oncotarget. 2013 Oct;4(10):1856-7.; Leary et al. Oncotarget. 2013 Aug;4(8):1119-20.; Leary et al. Sci Transl Med. 2012 Nov 28;4(162):162ra154.; Leary et al. Sci Transl Med. 2010 Feb 24;2(20):20ra14

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