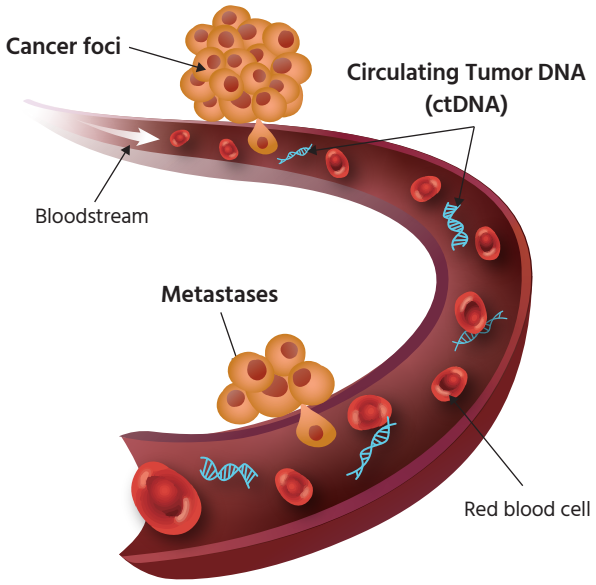
Pan-cancer test from a simple blood draw

Liquid Biopsy

What is the Liquid Biopsy?



Liquid Biopsy is an emerging technology developed to overcome the limitations of tissue biopsy and provide more options for patients and physicians batling cancer.

In cancer patients, **a fraction of the cfDNA is tumor derived.** These tumor derived DNA, called circulating tumor DNA(ctDNA), **carry the same genetic mutations present in the primary tumor cells.** From a single and simple blood drawing, with the right sensitivity and specificity, these can be used for diagnosis, treatment decisions and monitoring of cancers.

Test Subject

- Stage 3-4 Advanced, Metastatic and/or Recurrent solid tumor patients
- When tissue biopsy is not an option
- Carcinoma of Unknown Primary (CUP) patients

Compared to conventional tissue biopsy,

- You can have more comprehensive picture of tumor as tested tissue in tissue biopsy is just a portion of entire tumor.
- You can avoid delays in testing and the risk of complications caused by invasive tissue biopsies.
- It is a safe measure to alleviate the pain from repetitive tissue biopsies.

Gene List

Point mutation [46]	ALK	BRAF	BRCA1	BRCA2	EGFR	ERBB2	IDH1	IDH2	KIT	KRAS
	MYC	MYCN	NRAS	PDGFRA	AKT1	APC	AR	ARAF	CCND1	CCND2
	CCND3	CDK4	CDK6	CHEK2	CTNNB1	DDR2	ERBB3	ERG	ESR1	ETV1
	FBXW7	FGFR1	FGFR2	FGFR3	FGFR4	FLT3	GNA11	GNAQ	GNAS	HRAS
	MAP2K1	MAP2K2	MET	MTOR	NTRK1	NTRK3	PIK3CA	PTEN	RAF1	RET
	ROS1	SF3B1	SMAD4	SMO	TP53					
CNV [13]	EGFR	ERBB2	MYC	MYCN	CCND1	CCND2	CCND3	CDK4	CDK6	FGFR1
	FGFR2	FGFR3	MET							
RNA fusion [12]	ALK	BRAF	ERG	ETV1	FGFR1	FGFR2	FGFR3	MET	NTRK1	NTRK3
	RET	ROS1								

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Test Performance

	Reporting Threshold	Analytical Sensitivity			Analytical Specificity
SNVs	0.1* - 0.5% (2~3 molecules)	LOD	< 1%	92.1-96.9%	99.9%
			≥1%	100%	
Indel	0.1* - 0.5% (2~3 molecules)	LOD	< 1%	92.1-96.9%	99.9%
			≥1%	100%	
Fusions	1.0% (2 molecules)	100%			100%
CNVs	2.3 - 4.0 copies (gain) 1.0 copies (loss)	100%			100%

* Based on cfDNA input of 20 ng

Service features

Test	Oncomine Pan-cancer Cell-free (LBx) Assay	**Code**	O004
Specimen	Streck Tube WB 20 ml (10 ml X 2ea)	**TAT**	14 days
Method	NGS (Next Generation Sequencing)	**Sample Storage**	Room temperature (Refrigerated is recommended.)
Test description	As a test that analyzes genetic alteration of circulating tumor DNA by extracting cell-free DNA from a blood sample, it can help determine the treatment policy for malignant tumors. It can be applied to various malignant tumors such as stomach cancer, lung cancer, colon cancer, breast cancer, ovarian cancer, and prostate cancer. It can detect SNV, small indel, copy number variant, and gene rearrangement of 55 genes, and the detection limit of SNV and small indel mutations is about 0.1 to 2.0% depending on the input cfDNA concentration.		
Caution & Limitation	• This test was performed using DNA sequencing analysis, and it is possible to detect single nucleotide variant (SNV), small indel, copy number variation (CNV), gene rearrangement in the region included in the test. However it's not possible to detect any variants in the region not covered by the test. • The detection limit of SNV and small-indel depends on the amount of DNA in the extracted sample and limit of detection (LOD) is about 0.1~2.0% depending on the concentration of cfDNA. And when the tumor proportion is low, variants may not be detected, so clinical correlaton is recommended. • The four genes (APC, FBXW7, PTEN, TP53) corresponding to tumor suppressor genes among the selected genes included in this test do not cover the entire gene region, but include most of the major pathogenic variants. • This test does not distinguish germline and somatic variations. If the variant allele frequency of the mutation is close to 50% or 100% in the gene associated with hereditary cancer syndrome, there is a possibility of germline mutation. • The variants detected in this test are classified into four stages (tier 1 to 4) according to the 2017 JMD guideline (J Mol Diagn 2017; 19:313-327), and tier 4 variations are not reported.		

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