

Technical Specifications



About the Test

FoundationOne®CDx is a next generation sequencing based *in vitro* test for detection of substitutions, insertion and deletion alterations (indels), and copy number alterations (CNAs) in 324 genes and select gene rearrangements, as well as genomic signatures including microsatellite instability (MSI) and tumor mutational burden (TMB) using DNA isolated from formalin-fixed paraffin embedded (FFPE) tumor tissue specimens. The test can be used to identify patients who may benefit from treatment with targeted therapies in accordance with the approved therapeutic product labeling. Additionally, FoundationOne CDx is intended to provide tumor mutation profiling to be used by qualified health care professionals in accordance with professional guidelines in oncology for patients with solid malignant neoplasms. The FoundationOne CDx assay is a single-site assay performed at Foundation Medicine, Inc. in Cambridge, MA.



Performance Specifications

TECHNICAL INFORMATION	BASE SUBSTITUTIONS ¹	INDELS ¹	COPY NUMBER ALTERATIONS ¹	REARRANGEMENTS
Sensitivity	>99% MAF* ≥5%	>97% MAF* ≥10%	>95% CN ≥8 or 0 ≥30% tumour nuclei	≥90% ² >99% for ALK fusion ≥20% tumour nuclei
Specificity (PPV ^{**})	>99%	>99%	>99%	>99%
Typical Median depth of coverage (each covered read is of a unique DNA fragment to enable detection of alterations at low frequency)	500 ¹			
Sample requirements	≥40 μm tissue, of which a minimum of 20% is of malignant origin, on 8 to 10 unstained slides or in an FFPE block. Needle biopsy is also acceptable.			
Turn-around time	14 day average ^{***}			

* Mutant Allele Frequency as defined in Frampton *et al*

** Positive Predictive Value as defined in Frampton *et al*

*** As measured from the date the Foundation Medicine laboratory receives a sample that meets requirements.



Current Gene List[†]

Genes with full coding exonic regions included in FoundationOne CDx for the detection of substitutions, insertion-deletions (indels), and copy-number alterations (CNAs).

<i>ABL1</i>	<i>ACVR1B</i>	<i>AKT1</i>	<i>AKT2</i>	<i>AKT3</i>	<i>ALK</i>	<i>ALOX12B</i>	<i>AMER1 (FAM123B)APC</i>	
<i>AR</i>	<i>ARAF</i>	<i>ARFRP1</i>	<i>ARID1A</i>	<i>ASXL1</i>	<i>ATM</i>	<i>ATR</i>	<i>ATRX</i>	<i>AURKA</i>
<i>AURKB</i>	<i>AXIN1</i>	<i>AXL</i>	<i>BAP1</i>	<i>BARD1</i>	<i>BCL2</i>	<i>BCL2L1</i>	<i>BCL2L2</i>	<i>BCL6</i>
<i>BCOR</i>	<i>BCORL1</i>	<i>BRAF</i>	<i>BRCA1</i>	<i>BRCA2</i>	<i>BRD4</i>	<i>BRIP1</i>	<i>BTG1</i>	<i>BTG2</i>
<i>BTK</i>	<i>C11orf30 (EMSY)</i>	<i>CALR</i>	<i>CARD11</i>	<i>CASP8</i>	<i>CBFB</i>	<i>CBL</i>	<i>CCND1</i>	<i>CCND2</i>
<i>CCND3</i>	<i>CCNE1</i>	<i>CD22</i>	<i>CD274 (PD-L1)</i>	<i>CD70</i>	<i>CD79A</i>	<i>CD79B</i>	<i>CDC73</i>	<i>CDH1</i>
<i>CDK12</i>	<i>CDK4</i>	<i>CDK6</i>	<i>CDK8</i>	<i>CDKN1A</i>	<i>CDKN1B</i>	<i>CDKN2A</i>	<i>CDKN2B</i>	<i>CDKN2C</i>
<i>CEBPA</i>	<i>CHEK1</i>	<i>CHEK2</i>	<i>CIC</i>	<i>CREBBP</i>	<i>CRKL</i>	<i>CSF1R</i>	<i>CSF3R</i>	<i>CTCF</i>
<i>CTNNA1</i>	<i>CTNNB1</i>	<i>CUL3</i>	<i>CUL4A</i>	<i>CXCR4</i>	<i>CYP17A1</i>	<i>DAXX</i>	<i>DDR1</i>	<i>DDR2</i>
<i>DIS3</i>	<i>DNMT3A</i>	<i>DOTIL</i>	<i>EED</i>	<i>EGFR</i>	<i>EP300</i>	<i>EPHA3</i>	<i>EPHB1</i>	<i>EPHB4</i>
<i>ERBB2</i>	<i>ERBB3</i>	<i>ERBB4</i>	<i>ERCC4</i>	<i>ERG</i>	<i>ERRFI1</i>	<i>ESR1</i>	<i>EZH2</i>	<i>FAM46C</i>
<i>FANCA</i>	<i>FANCC</i>	<i>FANCG</i>	<i>FANCL</i>	<i>FAS</i>	<i>FBXW7</i>	<i>FGF10</i>	<i>FGF12</i>	<i>FGF14</i>
<i>FGF19</i>	<i>FGF23</i>	<i>FGF3</i>	<i>FGF4</i>	<i>FGF6</i>	<i>FGFR1</i>	<i>FGFR2</i>	<i>FGFR3</i>	<i>FGFR4</i>
<i>FH</i>	<i>FLCN</i>	<i>FLT1</i>	<i>FLT3</i>	<i>FOXL2</i>	<i>FUBP1</i>	<i>GABRA6</i>	<i>GATA3</i>	<i>GATA4</i>
<i>GATA6</i>	<i>GID4 (C17orf39)</i>	<i>GNA11</i>	<i>GNA13</i>	<i>GNAQ</i>	<i>GNAS</i>	<i>GRM3</i>	<i>GSK3B</i>	<i>H3F3A</i>
<i>HDAC1</i>	<i>HGF</i>	<i>HNF1A</i>	<i>HRAS</i>	<i>HSD3B1</i>	<i>ID3</i>	<i>IDH1</i>	<i>IDH2</i>	<i>IGF1R</i>
<i>IKBKE</i>	<i>IKZF1</i>	<i>INPP4B</i>	<i>IRF2</i>	<i>IRF4</i>	<i>IRS2</i>	<i>JAK1</i>	<i>JAK2</i>	<i>JAK3</i>

JUN	KDM5A	KDM5C	KDM6A	KDR	KEAP1	KEL	KIT	KLHL6
KMT2A (MLL)	KMT2D (MLL2)	KRAS	LTK	LYN	MAF	MAP2K1 (MEK1)	MAP2K2 (MEK2)	MAP2K4
MAP3K1	MAP3K13	MAPK1	MCL1	MDM2	MDM4	MEDI2	MEF2B	MEN1
MERTK	MET	MITF	MKNK1	MLH1	MPL	MRE11A	MSH2	MSH3
MSH6	MST1R	MTAP	MTOR	MUTYH	MYC	MYCL (MYCL1)	MYCN	MYD88
NBN	NF1	NF2	NFE2L2	NFKBIA	NKX2-1	NOTCH1	NOTCH2	NOTCH3
NPM1	NRAS	NT5C2	NTRK1	NTRK2	NTRK3	P2RY8	PALB2	PARK2
PARP1	PARP2	PARP3	PAX5	PBRM1	PDCD1 (PD-1)	PDCD1LG2 (PD-L2)	PDGFRA	PDGFRB
PKD1	PIK3C2B	PIK3C2G	PIK3CA	PIK3CB	PIK3R1	PIM1	PMS2	POLD1
POLE	PPARG	PPP2R1A	PPP2R2A	PRDM1	PRKAR1A	PRKCI	PTCH1	PTEN
PTPN11	PTPRO	QKI	RAC1	RAD21	RAD51	RAD51B	RAD51C	RAD51D
RAD52	RAD54L	RAF1	RARA	RB1	RBM10	REL	RET	RICTOR
RNF43	ROS1	RPTOR	SDHA	SDHB	SDHC	SDHD	SETD2	SF3B1
SGK1	SMAD2	SMAD4	SMARCA4	SMARCB1	SMO	SNCAIP	SOCS1	SOX2
SOX9	SPEN	SPOP	SRC	STAG2	STAT3	STK11	SUFU	SYK
TBX3	TEK	TET2	TGFBR2	TIPARP	TNFAIP3	TNFRSF14	TP53	TSC1
TSC2	TYRO3	U2AF1	VEGFA	VHL	WHSC1 (MMSET)	WHSC1L1	WT1	XPO1
XRCC2	ZNF217	ZNF703						

Select Rearrangements[†]

Genes with select intronic regions for the detection of gene rearrangements, one gene with a promoter region and one non-coding RNA gene.

ALK	BCL2	BCR	BRAF	BRCA1	BRCA2	CD74	EGFR	ETV4
ETV5	ETV6	EWSR1	EZR	FGFR1	FGFR2	FGFR3	KIT	KM2A (MLL)
MSH2	MYB	MYC	NOTCH2	NTRK1	NTRK2	NUTM1	PDGFRA	RAF1
RARA	RET	ROS1	RSPO2	SDC4	SLC34A2	TERC [‡]	TERT (promoter only) [§]	
TMPRSS2								

[†] TERC is non-coding RNA gene.

[§] TERT is gene with promoter region.

[‡] Current as of August 2019. Please visit foundationmedicine.com for the most up-to-date gene list.

References

1. Frampton *et al.* Development and validation of a clinical cancer genomic profiling test based on massively parallel DNA sequencing. *Nature Biotech* 2013 Nov.
2. Based on analysis of coverage and re-arrangement structure in the COSMIC database for solid tumour fusion genes where alteration prevalence could be established, complemented by detection of exemplar rearrangements in cell line titration experiments.
3. Based on ALK re-arrangement concordance analysis vs. a standard clinical FISH assay.

If you require this information in an accessible format, please contact Roche at 1-800-561-1759.

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