嘉義基督教醫院　新增基因檢測項目書面資料評估表

請購單號：N1120913024　申請單位：經營管理室　　申請人：汪妃芳　　分機：8512

廠商名稱: 賽亞基因科技股份有限公司 聯絡人: 蕭孟生 日期: 2023.09.05 聯絡方式: (02)89769123#7608

服務(醫令)項目名稱	FoundationOne®Liquid CDx 全方位癌症基因血液檢測[FoundationOne liquid-血液]				
1. 檢測標的基因清單	附件一_F1LCDx_ExUS_Technical Specifications_cc05				
2. 適用對象	針對所有沒有組織檢體的固態腫瘤患者				
3. 臨床意義/適應症	據研究，人體的兩萬個基因中，約有400個基因與癌症有高度的相關性，人體正常細胞中的這些癌症基因，發生先天或是後天的突變時，就會產生細胞不正常的增生，而可能導致癌症的發生。此產品使用血液檢體檢測ctDNA的突變狀態，並可檢測到>300個癌症相關基因，同時提供bTMB及MSI的評估報告，讓醫師能針對每個不同病人的狀況，進行個人化醫療，並做為免疫治療的參考。 附件二_F1L CDx Liquid Biopsy DA M-TW-00000309				
4.適用檢體種類	周邊全血 (8.5 mL x 2管)				
5.執行檢測之實驗室名稱及實驗室所在國	Foundation Medicine (美國)				
6. 檢驗方法	次世代定序 (NGS)				
7. 檢驗平台	Illumina NovaSeq 6000				
8. 針對 檢測標的基因 之試劑是否有IVD或FDA或其他核可(需提供佐證)	FDA核可 附件三_羅氏大藥廠FoundationMedicine US_FDA Approval Letter (F1LCDx)				
9. 實驗室針對檢測標的基因清單項目之認證$例如 CAP、TAF、LDTS(需提供佐證，例如證書、能力試驗結果...)	CAP認證 附件四(1)_[CAP認證]_2022_CAP_Accreditation_Certificate_CAM_exp 28Jan2025_Listing 附件四(2)_[CAP認證]_CAP certification_Cambridge_Activity Menu 附件四(3)_[能力試驗證明]_CFDNA-A_2022_LCDx				
10. 報告時效	30日曆天				
11.檢測效能*	偵測極限	sensitivity	specifity	陽性預測值	陰性預測值
(1)單核酸變異(snv)	Enhanced Sensitivity: 0.40% VAF ; Standard Sensitivity: 0.82% VAF	96.1%	>99.9%	無評估資料	無評估資料
(2)插入與缺失(Indels)	Enhanced Sensitivity: 0.40% VAF ; Standard Sensitivity: 0.82% VAF	100.0%	100.0%	無評估資料	無評估資料
(3)拷貝數變異(CNV)	Copy Number Losses: 30.4% TF ; Copy Number Amplification: 21.7% TF	無評估資料	無評估資料	無評估資料	無評估資料
(4)重組(rearrangements)	Enhanced Sensitivity: 0.37% VAF ; Standard Sensitivity is 0.90% VAF	100%	99.8%	無評估資料	無評估資料
(5) 融合(fusion)	無評估資料	無評估資料	無評估資	無評估資料	無評估資料
(6)蛋白質表現(protein expression)	NA	NA	NA	NA	NA
(7) 微衛星不穩定性檢測(MSI test)	0.8% Unstable loci	無評估資料	無評估資	無評估資料	無評估資料
■檢驗醫學科 □病理科填寫	評估意見	1.FoundationOne liquid-血液適用於所有沒有組織檢體的固態腫瘤患者之全方位癌症基因血液檢測，屬於特管法列管LDTS項目，送境外實驗室檢測 Foundation Medicine (美國)，須填"醫療機構施行實驗室開發檢測項目申請計畫書-境外實驗室適用"。 2.境外實驗室 Foundation Medicine，參與CAP認證，CAP認證編號:8057559，認證效期至114.01.28。				
	是否符合實驗室資料審查	■是 (審查條件: 提供品項已通過實驗室認證) □否 (審查條件: 提供品項未通過實驗室認證)			
	填表日期：20231023　填表人：李姵諭	單位主管：醫檢師 李姵諭 2023.10.23.	科室主任：檢驗科技術主任 賴美珠 2023.10.24		

說明: 無提供該項檢測填 NA；無該項評供資料則填:無評估資料

簽核流程：申請單位→檢驗醫學科/病理科→資材室採購組

修訂日期：109.06.18

Technical Specifications

FOUNDATIONONE®LIQUID CDx

Intended Use

FoundationOne Liquid CDx is a next generation sequencing based in vitro diagnostic device that analyzes 324 genes. Substitutions and insertion and deletion alterations (indels) are reported in 311 genes, copy number alterations (CNAs) are reported in 310 genes, and gene rearrangements are reported in 324 genes. The test also detects tumor fraction and the genomic signatures blood tumor mutational burden (bTMB) and microsatellite instability high (MSI-H) status. FoundationOne Liquid CDx utilizes circulating cell-free DNA (cfDNA) isolated from plasma derived from the anti-coagulated peripheral whole blood of cancer patients. The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with targeted therapies in accordance with the approved therapeutic product labeling. Additionally, FoundationOne Liquid 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 malignant neoplasms.

A negative result from a plasma specimen does not mean that the patient's tumor is negative for genomic findings. Patients who are negative for genomic findings should be reflexed to routine biopsy and their tumor mutation status confirmed using an FDA-approved tumor tissue test, if feasible.

FoundationOne Liquid CDx is a single-site assay performed at Foundation Medicine, Inc. in Cambridge, MA.

Summary of Analytical Sensitivity and Specificity

Results from our Limit of Detection (LoD) study are shown below, indicating the median variant allele frequency, tumor fraction or unstable loci at which the test has shown 95% probability of detection. Please refer to our product labeling for a list of the 75 genes baited for enhanced sensitivity and complete product specifications.

ALTERATION TYPE	BAIT SET REGION	MEDIAN LIMIT OF DETECTION (LOD)
Short Variants	Enhanced Sensitivity	0.40% VAF
	Standard Sensitivity	0.82% VAF
Rearrangements	Enhanced Sensitivity	0.37% VAF
	Standard Sensitivity	0.90% VAF
Copy Number Amplification	NA	21.7% TF
Copy Number Loss	NA	30.4% TF
MSI	NA	0.8% Unstable loci
bTMB (component indels)	NA	1.00% VAF
bTMB (component subs)	NA	1.00% VAF

VAF = variant allele frequency; TF = tumor fraction
The accuracy of %VAF / %TF have not been analytically validated

In our Limit of Blank study, which evaluated variant calling in healthy donors, 1,735 unique variants were included in the analysis for a total of 137,065 data points. A total of 18 false positives were observed across 4 unique short variants. The LoB was determined to be the ideal value of zero for short variants, rearrangements and CNAs. The false positive rate was shown to be 0% for rearrangements and CNAs and 0.013% (~1 in 8,000) for short variants (substitutions and indels).

FoundationOne Liquid CDx Gene List*

As a professional service, FoundationOne Liquid CDx interrogates 324 genes, including 309 genes with complete exonic (coding) coverage and 15 genes with only select non-coding coverage (indicated with an *); **75 genes (indicated in bold) are captured with increased sensitivity** and have complete exonic (coding) coverage unless otherwise noted. The test also detects tumor fraction and the genomic signatures blood mutational burden (bTMB) and microsatellite instability high (MSI-H) status.

ABL1 ***[Exons 4-9]***
ACVR1B
AKT1 ***[Exon 3]***
AKT2
AKT3
ALK ***[Exons 20-29 Introns 18,19]***
ALOX12B
AMER1 (FAM123B)
APC
AR
ARAF ***[Exons 4, 5, 7, 11, 13, 15, 16]***
ARFRP1
ARID1A
ASXL1
ATM
ATR
ATRX
AURKA
AURKB
AXIN1
AXL
BAP1
BARD1
BCL2
BCL2L1
BCL2L2
BCL6
BCOR
BCORL1
*BCR** *[Introns 8, 13, 14]*
BRAF ***[Exons 11-18,*** *Introns 7-10]*
BRCA1 *[Introns 2, 7, 8, 12, 16, 19, 20]*
BRCA2 *[Intron 2]*
BRD4
BRIP1
BTG1
BTG2
BTK ***[Exons 2, 15]***
C11orf30 (EMSY)
C17orf39 (GID4)
CALR
CARD11
CASP8
CBFB
CBL
CCND1
CCND2
CCND3
CCNE1
CD22

FoundationOne Liquid CDx Gene List continued →

(FoundationOne Liquid CDx Gene List continued)

CD70

*CD74**
[Introns 6-8]

CD79A

CD79B

CD274 (PD-L1)

CDC73

CDH1

CDK12

CDK4

CDK6

CDK8

CDKN1A

CDKN1B

CDKN2A

CDKN2B

CDKN2C

CEBPA

CHEK1

CHEK2

CIC

CREBBP

CRKL

CSF1R

CSF3R

CTCF

CTNNA1

CTNNB1
[Exon 3]

CUL3

CUL4A

CXCR4

CYP17A1

DAXX

DDR1

DDR2
[Exons 5, 17, 18]

DIS3

DNMT3A

DOT1L

EED

EGFR
[Introns 7, 15, 24-27]

EP300

EPHA3

EPHB1

EPHB4

ERBB2

ERBB3
[Exons 3, 6, 7, 8, 10, 12, 20, 21, 23, 24, 25]

ERBB4

ERCC4

ERG

ERRFI1

ESR1
[Exons 4-8]

*ETV4**
[Intron 8]

*ETV5**
[Introns 6,7]

ETV6*
[Introns 5,6]

*EWSR1**
[Introns 7-13]

EZH2
[Exons 4, 16, 17, 18]

*EZR**
[Introns 9-11]

FAM46C

FANCA

FANCC

FANCG

FANCL

FAS

FBXW7

FGF10

FGF12

FGF14

FGF19

FGF23

FGF3

FGF4

FGF6

FGFR1
[Introns 1, 5, ***Intron 17]***

FGFR2
[Intron 1, ***Intron 17]***

FGFR3
[Exons 7, 9 (alternative designation exon 10), 14, 18, Intron 17]

FGFR4

FH

FLCN

FLT1

FLT3
[Exons 14, 15, 20]

FOXL2

FUBP1

GABRA6

GATA3

GATA4

GATA6

GNA11
[Exons 4, 5]

GNA13

GNAQ
[Exons 4, 5]

GNAS
[Exons 1, 8]

GRM3

GSK3B

H3F3A

HDAC1

HGF

HNF1A

HRAS
[Exons 2, 3]

HSD3B1

ID3

IDH1
[Exon 4]

IDH2
[Exon 4]

IGF1R

IKBKE

IKZF1

INPP4B

IRF2

IRF4

IRS2

JAK1

JAK2
[Exon 14]

JAK3
[Exons 5, 11, 12, 13, 15, 16]

JUN

KDM5A

KDM5C

KDM6A

KDR

KEAP1

KEL

KIT
[Exons 8, 9, 11, 12, 13, 17, *Intron 16]*

KLHL6

KMT2A (MLL)
[Introns 6, 8-11, ***Intron 7]***

KMT2D (MLL2)

KRAS

LTK

LYN

MAF

MAP2K1 (MEK1)
[Exons 2, 3]

MAP2K2 (MEK2)
[Exons 2-4, 6, 7]

MAP2K4

MAP3K1

MAP3K13

MAPK1

MCL1

MDM2

MDM4

MED12

MEF2B

MEN1

MERTK

MET

MITF

MKNK1

MLH1

MPL
[Exon 10]

MRE11A

MSH2
[Intron 5]

MSH3

MSH6

MST1R

MTAP

MTOR
[Exons 19, 30, 39, 40, 43-45, 47, 48, 53, 56]

MUTYH

*MYB**
[Intron 14]

MYC
[Intron 1]

MYCL (MYCL1)

MYCN

MYD88
[Exon 4]

NBN

NF1

NF2

NFE2L2

NFKBIA

NKX2-1

NOTCH1

NOTCH2
[Intron 26]

NOTCH3

NPM1
[Exons 4-6, 8, 10]

NRAS
[Exons 2, 3]

NSD3 (WHSC1L1)

NT5C2

NTRK1
[Exons 14, 15, Introns 8-11]

NTRK2
[Intron 12]

NTRK3
[Exons 16, 17]

*NUTM1**
[Intron 1]

P2RY8

PALB2

PARK2

PARP1

PARP2

PARP3

PAX5

PBRM1

PDCD1 (PD-1)

PDCD1LG2 (PD-L2)

PDGFRA
[Exons 12, 18, Introns 7, 9, 11]

PDGFRB
[Exons 12-21, 23]

PDK1

PIK3C2B

PIK3C2G

PIK3CA
Exons 2, 3, 5-8, 10, 14, 19, 21 (Coding Exons 1, 2, 4-7, 9, 13, 18, 20)

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
[Exons 3, 4, 6, 7, 10, 14, 15, 17, *Introns 4-8]*

RARA
[Intron 2]

RB1

RBM10

REL

RET
[Introns 7, 8, ***Exons 11, 13-16, Introns 9-11]***

RICTOR

RNF43

ROS1
[Exons 31, 36-38, 40, Introns 31-35]

RPTOR

*RSPO2**
[Intron 1]

*SDC4**
[Intron 2]

SDHA

SDHB

SDHC

SDHD

SETD2

SF3B1

SGK1

*SLC34A2**
[Intron 4]

SMAD2

SMAD4

SMARCA4

SMARCB1

SMO

SNCAIP

SOCS1

SOX2

SOX9

SPEN

SPOP

SRC

STAG2

STAT3

STK11

SUFU

SYK

TBX3

TEK

TERC {ncRNA}*

TERT* {Promoter}

TET2

TGFBR2

TIPARP

*TMPRSS2**
[Introns 1-3]

TNFAIP3

TNFRSF14

TP53

TSC1

TSC2

TYRO3

U2AF1

VEGFA

VHL

WHSC1

WT1

XPO1

XRCC2

ZNF217

ZNF703

Visit foundationmedicine.com to create an online account.
†Current as of August 2020. Please visit foundationmedicine.com for the most up-to-date gene list.

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COLLEGE of AMERICAN PATHOLOGISTS

CERTIFICATE OF ACCREDITATION

Foundation Medicine Laboratory
Cambridge, Massachusetts
Julia A. Elvin, MD, PhD

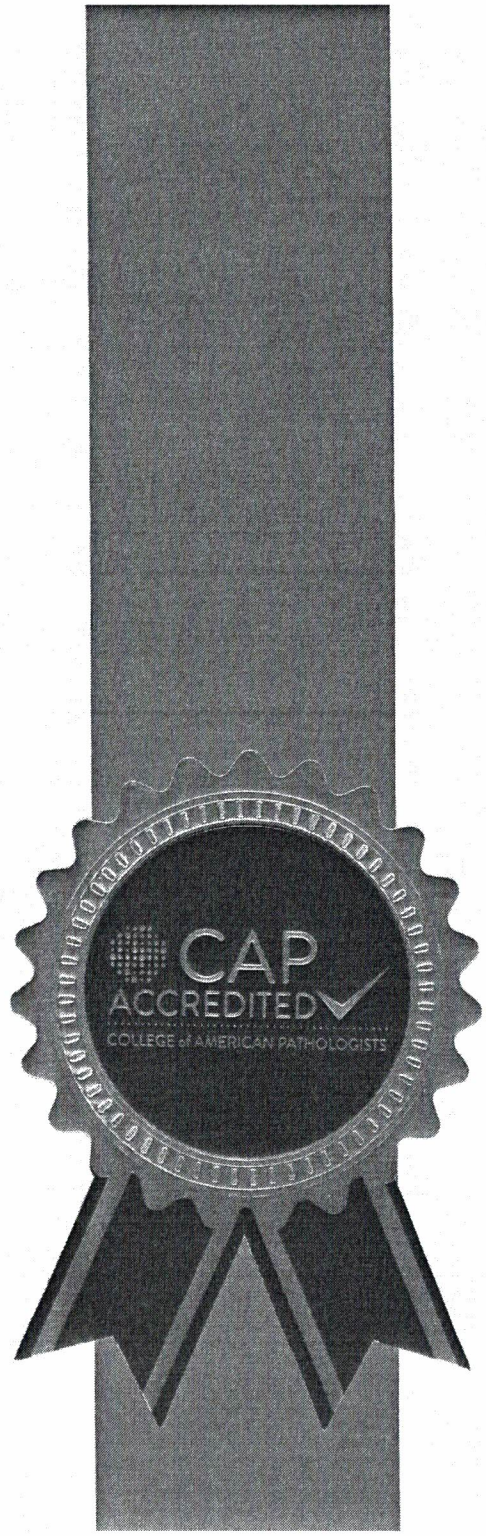
CAP Number: 8057559
AU-ID: 1656274
CLIA Number: 22D2027531

The organization named above meets all applicable standards for accreditation and is hereby accredited by the College of American Pathologists' Laboratory Accreditation Program. Reinspection should occur prior to **January 28, 2025** to maintain accreditation.

Accreditation does not automatically survive a change in director, ownership, or location and assumes that all interim requirements are met.

Kathleen G. Beavis, MD, Accreditation Committee Chair

Emily Volk, MD, FCAP, President, College of American Pathologists



Foundation Medicine Laboratory (CAP# 8057559)

Anatomic Pathology (Section ID: 2074382) - Activities

Activity Name	Discipline	Subdiscipline	Type
Microscopic evaluation, Surgical Pathology	Anatomic Pathology	Surgical Pathology	S
Routine processing (e.g., block prep, H&E stain)	Anatomic Pathology	Anatomic Pathology Processing	S
Specimen adequacy, microscopic (at collection)	Anatomic Pathology	Anatomic Pathology Processing	S

Foundation Medicine Laboratory (CAP# 8057559)

Foundation Medicine Laboratory (Section ID: 1661908) - Activities

Activity Name	Discipline	Subdiscipline	Type
Copy number variant analysis, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	R
Copy number variant analysis, Molecular Pathology	Molecular Pathology	Molecular Pathology	R
Copy number variant analysis, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	R
Laboratory developed test (LDT), Hematologic	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
Laboratory developed test (LDT), Molecular Path	Molecular Pathology	Molecular Pathology	S
Laboratory developed test (LDT), Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
Microsatellite instability (MSI)	Molecular Pathology	Molecular Oncology - Solid Tumor	R
Molecular analysis of leukemias and lymphomas	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
Molecular oncology assay, other, Hematologic	Molecular Pathology	Molecular Oncology - Hematologic Diseases	R
Molecular oncology assay, other, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	R
Neoplastic cell content, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
Neoplastic cell content, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
NGS, analytical wet bench, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
NGS, analytical wet bench, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
NGS, analytical wet bench, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
NGS, bioinformatics, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
NGS, bioinformatics, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
NGS, bioinformatics, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
NGS, interpretation, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
NGS, interpretation, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
NGS, interpretation, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
NGS, panel sequencing, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	R
NGS, panel sequencing, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
NGS, panel sequencing, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	R
Non-invasive circulating tumor cfDNA / cfRNA	Molecular Pathology	Molecular Oncology - Solid Tumor	R
Nucleic acid amplification, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
Nucleic acid amplification, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
Nucleic acid amplification, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
Nucleic acid extraction, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S

Foundation Medicine Laboratory (CAP# 8057559)

Foundation Medicine Laboratory (Section ID: 1661908) - Activities

Activity Name	Discipline	Subdiscipline	Type
Nucleic acid extraction, Molecular Pathology	Molecular Pathology	Molecular Pathology	S
Nucleic acid extraction, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S
RNA sequencing, Hematologic Diseases	Molecular Pathology	Molecular Oncology - Hematologic Diseases	S
RNA sequencing, Solid Tumor	Molecular Pathology	Molecular Oncology - Solid Tumor	S