



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: N1120925002 申請單位: 血液腫瘤科 申請人: 王毓楷 分機: 8521
 廠商名稱: 艾爾瑪生技股份有限公司 聯絡人: 王仁中 日期: 20230918 聯絡方式: 0970-537-591

服務(醫令)項目名稱		Invitae 遺傳性癌症全方位基因檢測				
1. 檢測標的基因清單		AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, CTNNA1, DICER1, DIS3L2, EGFR, EPCAM, FH, FLCN, GATA2, GPC3, GREM1, HOXB13, HRAS, KIT, MAX, MEN1, MET, MTF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERC, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1				
2. 適用對象		<p style="color: red; font-weight: bold;">限三陰性乳癌第III期或第IV期病人可開立, 右符合輝瑞藥廠贊助檢測費</p> <ul style="list-style-type: none"> ●本人或血親為早期癌症患者: 小於50歲發病確診罹患癌症。 ●本人或血親患有以下癌症: 乳癌(尤其三陰性、男性乳癌)、婦癌(尤其卵巢癌、子宮內膜癌)、攝護腺癌、大腸直腸癌、小腸癌、胰臟癌、胃癌、甲狀腺髓質癌、泌尿道癌、腦瘤、膽道癌。 ●本人或血親被診斷出兩種以上不同癌症: 人體2個不同區域的癌症, 或同一器官中2個單獨的癌症。 ●血親家族中有多個親戚患有相同或相關類型癌症。 ●大腸鏡中發現10個以上的息肉。 ●血親中曾有被檢測出具有增加癌症風險的基因變異。 				
3. 臨床意義/適應症		<p>施作本檢測可了解受測者是否有罹患某些疾病的高風險或遺傳風險, 更主動地關心健康, 並做出正確的醫療決定, 以降低風險。</p> <p>此檢測有再確認機制, BRCA 有約 10% 為大片斷重組 (Large rearrangement), 這種變異NGS 一般無法偵測, Invitae 會以 MLPA (Gold standard) 去做確認, 確保所有 BRCA 突變都能被找出(https://www.invitae.com/en/provider_faqs/tech_and_quality)。</p>				
4. 適用檢體種類		血液				
5. 執行檢測之實驗室名稱及實驗室所在國別		Invitae Corporation Laboratory (1400 16th St, San Francisco, CA, USA 94103)				
6. 檢驗方法		次世代定序 (Next Generation Sequencing, NGS), Illumina				
7. 檢驗平台		採用先進的次世代定序技術, 從提交的檢體中獲得的基因組DNA使用PCR-free方法進行定序(針對整個基因組進行定序)。對於不符合Invitae次世代定序嚴格質量指標的臨床發現, 將使用下列正交方法進行確認, 如: PacBio定序、Sanger定序、比較基因組雜交 (aCGH) 和多重連接依賴性探針擴增 (MLPA)。(將依據變異情形使用對應的檢測方法。)				
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)		無				
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...))		(請見附件)				
10. 報告時效		報告無時效限制				
11. 檢測效能*		偵測極限	sensitivity	specificity	陽性預測值	陰性預測值
(1)單核酸變異(snv)		N/A	>99%	>99%	N/A	N/A
(2)插入與缺失(Indels)		N/A	>99%	>99%	N/A	N/A
(3)拷貝數變異(CNV)		N/A	>99%	>99%	N/A	N/A
(4)重組(rearrangements)		N/A	N/A	N/A	N/A	N/A
(5)融合(fusion)		N/A	N/A	N/A	N/A	N/A
(6)蛋白質表現(protein expression)		N/A	N/A	N/A	N/A	N/A
(7) 微衛星不穩定性檢測(MSI test)		N/A	N/A	N/A	N/A	N/A
		N/A: Not applicable as these terms are meant for tumor/somatic tissue/liquid NGS tests.				
■ 檢驗醫學科	評估意見	1. 境外實驗室Invitae Corporation Laboratory 為CAP認證實驗室, 編號8770489, 認證期限至2024.10.03, 符合規定。 2. 屬於特管法列管LDTs項目, 送境外實驗室檢測 Invitae Corporation Laboratory (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"。				
□ 病理科填寫	是否符合實驗室資料審查	<input checked="" type="checkbox"/> 是 (審查條件: 提供品項已通過實驗室認證) <input type="checkbox"/> 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: 李嫻諭 單位主管: 科主任:						

說明: 無提供該項檢測填 NA; 無該項評估資料則填: 無評估資料

審核流程: 申請單位→檢驗醫學科/病理科→資料室採購組

修訂日期: 109.06.18



COLLEGE of AMERICAN
PATHOLOGISTS

CERTIFICATE OF ACCREDITATION

**Invitae Corporation
Laboratory
San Francisco, California
Kristin Gibson, PhD, ACMG**

CAP Number: 8770489
AU-ID: 1703789
CLIA Number: 05D2040778

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 **October 3, 2024** to maintain accreditation.

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

Michael Bradley Datto, MD, PhD, FCAP
Chair, Accreditation Committee

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



San Francisco Section_Molecular_Genetics(1719886)_Activities.04_10_2023_2_47

Invitae Corporation (CAP# 8770489)			
Molecular Genetics (Section ID: 1719886) - Activities			
Activity Name	Discipline	Subdiscipline	Type
Genomic Copy Number - Microarray, Inherited	Molecular Pathology	Inherited Genetics	R
Alpha-thalassemia	Molecular Pathology	Inherited Genetics	R
Usher syndrome	Molecular Pathology	Inherited Genetics	R
Polycystic kidney disease	Molecular Pathology	Inherited Genetics	R
Arrays &/or Microarrays, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Non-invasive prenatal testing (NIPT)	Molecular Pathology	Inherited Genetics	R
NGS, panel sequencing, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Methylenetetrahydrofolate reductase (MTHFR)	Molecular Pathology	Inherited Genetics	R
Laboratory developed test (LDT),Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Severe combined immunodeficiency (SCID)	Molecular Pathology	Inherited Genetics	R
NGS, interpretation, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
NGS, bioinformatics, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
NGS, analytical wet bench, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Whole exome sequencing, Inherited Genetics	Molecular Pathology	Inherited Genetics	R
Mucopolipidosis type IV	Molecular Pathology	Inherited Genetics	R
Phenylketonuria (PKU)	Molecular Pathology	Inherited Genetics	R
Zellweger syndrome	Molecular Pathology	Inherited Genetics	R
Treacher Collins syndrome	Molecular Pathology	Inherited Genetics	R
Noonan syndrome	Molecular Pathology	Inherited Genetics	R
Mucopolysaccharidosis	Molecular Pathology	Inherited Genetics	R
Multiple endocrine neoplasia 1 (MEN1)	Molecular Pathology	Inherited Genetics	R
Von Hippel-Lindau	Molecular Pathology	Inherited Genetics	R
Saethre-Chotzen syndrome	Molecular Pathology	Inherited Genetics	R
Opitz G/BBB syndrome	Molecular Pathology	Inherited Genetics	R
Rett syndrome	Molecular Pathology	Inherited Genetics	R

Molecular genetics assay, other	Molecular Pathology	Inherited Genetics	R
Prothrombin (F2)	Molecular Pathology	Inherited Genetics	R
Nucleic acid extraction, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Niemann-Pick disease	Molecular Pathology	Inherited Genetics	R
Tay-Sachs disease	Molecular Pathology	Inherited Genetics	R
Spinal muscular atrophy	Molecular Pathology	Inherited Genetics	R
Multiple endocrine neoplasia 2 (MEN2)	Molecular Pathology	Inherited Genetics	R
Nucleic acid amplification, Inherited Genetics	Molecular Pathology	Inherited Genetics	S
Loeys-Dietz syndrome (LDS)	Molecular Pathology	Inherited Genetics	R
Joubert syndrome	Molecular Pathology	Inherited Genetics	R
Familial mediterranean fever	Molecular Pathology	Inherited Genetics	R
Charcot-Marie-Tooth disease	Molecular Pathology	Inherited Genetics	R
Maple syrup urine disease	Molecular Pathology	Inherited Genetics	R
Familial dysautonomia	Molecular Pathology	Inherited Genetics	R
Bloom syndrome	Molecular Pathology	Inherited Genetics	R
Medium-chain acyl-coA dehydrogenase (MCAD)	Molecular Pathology	Inherited Genetics	R
Fabry disease	Molecular Pathology	Inherited Genetics	R
Marfan syndrome	Molecular Pathology	Inherited Genetics	R
Long QT syndrome	Molecular Pathology	Inherited Genetics	R
Glycogen storage disease type IA	Molecular Pathology	Inherited Genetics	R
Lynch syndrome	Molecular Pathology	Inherited Genetics	R
Familial adenomatous polyposis	Molecular Pathology	Inherited Genetics	R
Collagen (e.g., COL2A1, COL11A1)	Molecular Pathology	Inherited Genetics	R
Galactosemia	Molecular Pathology	Inherited Genetics	R
ARX related disorders	Molecular Pathology	Inherited Genetics	R
Hereditary paraganglioma-pheochromocytoma	Molecular Pathology	Inherited Genetics	R
Hereditary papillary renal carcinoma	Molecular Pathology	Inherited Genetics	R
Crouzon syndrome	Molecular Pathology	Inherited Genetics	R
CHARGE syndrome	Molecular Pathology	Inherited Genetics	R
Birt-Hogg-Dube	Molecular Pathology	Inherited Genetics	R
Apert syndrome	Molecular Pathology	Inherited Genetics	R
Factor V Leiden	Molecular Pathology	Inherited Genetics	R
DNA sequencing (Sanger, Pyro), Inherited Genetics	Molecular Pathology	Inherited Genetics	S

Fanconi Anemia	Molecular Pathology	Inherited Genetics	R
Canavan Disease	Molecular Pathology	Inherited Genetics	R
Hemochromatosis	Molecular Pathology	Inherited Genetics	R
Fragile X	Molecular Pathology	Inherited Genetics	R
DMD/Becker	Molecular Pathology	Inherited Genetics	R
Cystic fibrosis	Molecular Pathology	Inherited Genetics	R
BRAF, Inherited Genetics	Molecular Pathology	Inherited Genetics	R
EGFR, Inherited Genetics	Molecular Pathology	Inherited Genetics	R
KRAS, Inherited Genetics	Molecular Pathology	Inherited Genetics	R

San Francisco Lab College of American Pathologists (CAP) Proficiency Testing Program

CAP Survey	Description	Genes
NGS	NGS - Germline	200 Genes that varies each round
MGL1	CAP/ACMG Molecular Genetics	F2, F5, FMR1, HFE
MGL2	CAP/ACMG Molecular Genetics	DMD, MTHFR, SMN1, SMN2, CFTR
MGL3	CAP/ACMG Molecular Genetics	BRCA1, BRCA2, GJB2, RET
MGL4	CAP/ACMG Molecular Genetics	ASPA, HEXA, ELP1, BLM, SMP1, GBA, G6PC, MCOLN1, FANCC
MGL5	CAP/ACMG Molecular Genetics	CFTR
NIPT	Non-Invasive Prenatal Testing	N/A
IMD	IMD2 / MCAD	ACADM
RETT	Rett Syndrome	MECP2



**COLLEGE of AMERICAN
PATHOLOGISTS**

325 Waukegan Road, Northfield, Illinois 60093-2750
800-323-4040 • cap.org

ORIGINAL EVALUATION

MGL3-B 2022 Molecular Genetics Series

INSTITUTION: Invitae Corporation
San Francisco CA 94103-5110

ATTENTION: Gicelle Arquero

CAP NUMBER: 8770489-03 **Kit#** 1

KIT INFORMATION:	Kit ID:	Kit Mailed:	Original Evaluation:	Next Mailing Date:
	35691128	11/21/2022	1/19/2023	6/12/2023

COPIED TO: LAP CMS (05D2040778) NYS Dept of Health CLEP (8884)

IMPORTANT NOTES: For all homozygous acceptable genotype results listed on your evaluation, only one allele can be shown as reported by your laboratory due to restrictions with our database.

This should not affect the grading of your genotyping data. Complete genotyping results are available in the MGL3 Participant Summary.

LEGEND: Exception Reason Codes appearing in this evaluation:
<NONE>

Swaroop Aradhya, Ph.D

Signed Electronically

Reviewed By

Date



**COLLEGE of AMERICAN
PATHOLOGISTS**

325 Waukegan Road, Northfield, Illinois 60093-2750
800-323-4040 • cap.org

CAP Number: 8770489-03 **Kit#** 1
Institution: Invitae Corporation
Attention: Gicelle Arquero
City / State: San Francisco CA 94103-5110

Kit ID: 35691128
Kit Mailed: 11/21/2022
Original Evaluation: 1/19/2023

**EVALUATION
ORIGINAL**

MGL3-B 2022 Molecular Genetics Series

Test Method	Specimen	Your Result	Intended Response	Your Grade
BRCA1/2 Allele 1 & 2 NEXT-GEN SEQUENCING LAB-DEVELOPED TEST	MGL3-04	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
	MGL3-05	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
	MGL3-06	BRCA1 c.68_69del NO VARIANT DETECTED	BRCA1 c.68_69del NO VARIANT DETECTED	Good
BRCA1/2 Interpretation	MGL3-04	RISK CANT BE DETERMIND	RISK CANT BE DETERMIND	Good
	MGL3-05	RISK CANT BE DETERMIND	RISK CANT BE DETERMIND	Good
	MGL3-06	RISK ABOVE GENL POPUL	RISK ABOVE GENL POPUL	Good
Connexin Genotype-AL 1 NEXT-GEN SEQUENCING LAB-DEVELOPED TEST	MGL3-04	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
Connexin Genotype-AL 1 NEXT-GEN SEQUENCING LAB-DEVELOPED TEST	MGL3-05	PATHOGENIC VARIANT NO VARIANT DETECTED	PATHOGENIC VARIANT NO VARIANT DETECTED	Good
Connexin 26 Allele 1 &	MGL3-05	c.35del, p.Gly12Valfs	c.35del, p.Gly12Valfs	Good
Connexin Genotype-AL 1 NEXT-GEN SEQUENCING LAB-DEVELOPED TEST	MGL3-06	NO VARIANT DETECTED	NO VARIANT DETECTED	Good

The College of American Pathologists recommends that the result of this interlaboratory comparison not be used as a sole criterion for judging the performance of any individual clinical laboratory.



**COLLEGE of AMERICAN
PATHOLOGISTS**

325 Waukegan Road, Northfield, Illinois 60093-2750
800-323-4040 • cap.org

CAP Number: 8770489-03 **Kit#** 1
Institution: Invitae Corporation
Attention: Gicelle Arquero
City / State: San Francisco CA 94103-5110

Kit ID: 35691128
Kit Mailed: 11/21/2022
Original Evaluation: 1/19/2023

**EVALUATION
ORIGINAL**

MGL3-B 2022 Molecular Genetics Series

Test Method	Specimen	Your Result	Intended Response	Your Grade
Connexin 26 Interpretation	MGL3-04	NEG FOR DFNB1 VARIANTS	NEG FOR DFNB1 VARIANTS	Good
	MGL3-05	PT IS CARRIER OF DFNB1	PT IS CARRIER OF DFNB1	Good
	MGL3-06	NEG FOR DFNB1 VARIANTS	NEG FOR DFNB1 VARIANTS	Good
MEN2 Genotype NEXT-GEN SEQUENCING LAB-DEVELOPED TEST	MGL3-04	p.Cys620Phe	p.Cys620Phe	Good
	MGL3-05	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
	MGL3-06	NO VARIANT DETECTED	NO VARIANT DETECTED	Good
MEN2 Interpretation	MGL3-04	PT DX MEN2A OR MTC	PT DX MEN2A OR MTC	Good
	MGL3-05	UNLIKELY HERITABLE DIS	UNLIKELY HERITABLE DIS	Good
	MGL3-06	UNLIKELY HERITABLE DIS	UNLIKELY HERITABLE DIS	Good

The College of American Pathologists recommends that the result of this interlaboratory comparison not be used as a sole criterion for judging the performance of any individual clinical laboratory.