LifeCell Diagnostics is now mfine /diagnostics

Mammalife Panels

Trusted by Clinicians for Screening & Diagnosis of Breast Cancer

Breast Cancer Markers

To predict the prognosis of breast cancer and to guide its therapy

ER/PR

Hormonal Therapy

Her2

Herceptin, Poorer Prognosis

Ki 67 & p53

Proliferation Index

Neo Adjuvant Therapy & Closer Follow Up

Genetic Testing for Hereditary Breast & Ovarian Cancer

The American Society of Breast Surgeons recommends genetic testing to be made available to all breast cancer patients

Breast cancer risk is associated with many factors, genetic predisposition with mutations in inherited genes accounts for

10%

of all breast cancers

Awareness, pre-emptive testing and risk management is the KEY

While **BRCA1** and **BRCA2** mutations are the most common causes, other genes such as

CDH1 | BARD1 | CHEK2 | PALB2 | PTEN | TP53

etc.

Can also contribute to the risk of breast and ovarian cancer

With proficiency in genetic testing, LifeCell offers the most comprehensive & up-to-date panels:

New Launch

ONCO0065 : OncoLife-Homologous Recombination Repair (HRR) 27 genes Panel

Clinical utility: Offers important value addition to the patient by analysing mutations in Homologous Recombinant Repair pathway genes which are predictive biomarkers for PARP inhibitor therapy

Genes Covered: 27 Genes

Platform: Next-Generation (NGS) on Illumina NovaSeq 6000

Assay Detects: SNVs, Indels & Copy number variations

Read depth: >250x

Highly Penetrant Genes:

BRCA1, BRCA2, CDK12, FANCL, NBN, PPP2R2A, PTEN, RAD51B, RAD51C, RAD51D, RAD54L,TP53

Moderately Penetrant Genes:

ATM, BARD1, BRIP1, CHEK2, FANCD2, KRAS, MRE11, PIK3CA,POLD1, POLE, RAD50, RAD51, RAD52, XRCC2

GEN0487O : Hereditary cancer gene panel

Clinical utility: The test analyzes genes associated with Hereditary Cancers

Genes Covered: 106 Genes

Platform: Whole Exome sequencing

Assay Detects: Missense / Nonsense / Splice Site / Inframe Deletions / Frameshift / CNV Coverage

Read depth: 80-100x

Also available

GEN0357O	B0059	B0059a
BRCA1 & BRCA2 Gene analysis	BRCA1 & BRCA2 Deletion/Duplication analysis by MLPA	BRCA1 & BRCA2 Mutation analysis

GENES COVERED

GENE NAME	CANCER
AIP	Pituitary Adenoma
ALK	Neuroblastoma
APC	Colorectal cancer
AR	Prostate cancer
ATM	Breast cancer,
BAP1	Tumor predisposition syndrome 1
BARD1	Breast cancer, susceptibility
BLM	Bloom syndrome
BMPR1A	Polyposis syndrome
BRCA1	Breast-ovarian cancer, familial,
BRCA2	Breast-ovarian cancer,
BRIP1	Breast cancer, early-onset, susceptibility to
BUB1B	Colorectal cancer, somatic
CD82	Prostate cancer
CDC73	Parathyroid carcinoma
CDH1	Endometrial carcinoma, somatic
CDK4	Melanoma, cutaneous malignant, 3
CDKN1C	Beckwith-Wiedemann syndrome
CDKN2A	Melanoma, cutaneous malignant, 2
CEBPA	Leukemia, acute myeloid, somatic
CEP57	Mosaic variegated aneuploidy syndrome 2
CHEK2	Osteosarcoma, somatic
CYLD	Brooke-Spiegler syndrome
DDB2	Xeroderma pigmentosum, group E, DDB-negative subtype
DICER1	GLOW syndrome, somatic mosaic
DIS3L2	Perlman syndrome
EGFR	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in
ELAC2	Prostate cancer, hereditary, 2, susceptibility to
ENG	Telangiectasia, hereditary hemorrhagic, type 1
EPCAM	Colorectal cancer, hereditary nonpolyposis, type 8
ERCC2	Trichothiodystrophy 1, photosensitive
ERCC3	Trichothiodystrophy 2, photosensitive
ERCC4	Xeroderma pigmentosum, group F
ERCC5	Xeroderma pigmentosum, group G
EXT1	Exostoses, multiple, type 1
EXT2	Exostoses, multiple, type 2
EZH2	Weaver syndrome
FANCA	Fanconi anemia, complementation group A
FANCB	Fanconi anemia, complementation group B

GENE NAME	CANCER
FANCC	Fanconi anemia, complementation group C
FANCD2	Fanconi anemia, complementation group D2
FANCE	Fanconi anemia, complementation group E
FANCF	Fanconi anemia, complementation group F
FANCG	Fanconi anemia, complementation group G
FANCI	Fanconi anemia, complementation group I
FANCL	Fanconi anemia, complementation group L
FANCM	Premature ovarian failure 15
FH	Leiomyomatosis and renal cell cancer
FLCN	Colorectal cancer, somatic
GATA2	Leukemia, acute myeloid, susceptibility to
GPC3	Wilms tumor, somatic
HRAS	Thyroid carcinoma, follicular, somatic
KIT	Gastrointestinal stromal tumor, familial
MAX	Pheochromocytoma, susceptibility to
MEN1	Carcinoid tumor of lung
MET	Hepatocellular carcinoma, childhood type, somatic
MLH1	Colorectal cancer, hereditary nonpolyposis, type 2
MLH3	Colorectal cancer, hereditary nonpolyposis, type 7
MRE11A	Ataxia-telangiectasia-like disorder 1
MSH2	Colorectal cancer, hereditary nonpolyposis, type 1
MSH3	Familial adenomatous polyposis 4/ Endometrial carcinoma, somatic
MSH6	Colorectal cancer, hereditary nonpolyposis, type 5
MSR1	Barrett esophagus/esophageal adenocarcinoma
MUTYH	Adenomas, multiple colorectal
MXI1	Prostate cancer, somatic
NBN	Leukemia, acute lymphoblastic
NF1	Leukemia, juvenile myelomonocytic/Neurofibromatosi, type 1
NF2	Meningioma, NF2-related, somatic/Neurofibromatosis, type 2
NSD1	Sotos syndrome
PALB2	breast cancer/pancreatic cancer
PHOX2B	Neuroblastoma with Hirschsprung disease
PMS1	hereditary nonpolyposis colon cancer
PMS2	Colorectal cancer, hereditary nonpolyposis, type 4

GENE NAME	CANCER
PRF1	Lymphoma, non-Hodgkin
PRKAR1A	Adrenocortical tumor, somatic
PTCH1	Holoprosencephaly 7
PTEN	Prostate cancer/menangioma
RAD5	
RAD51C	Breast-ovarian cancer, familial, susceptibility to, 3
RAD51D	Breast-ovarian cancer, familial, susceptibility to, 4
RB1	Retinoblastoma
RECQL4	Rothmund-Thomson syndrome, type 2/Baller-Gerold syndrome/RAPADILINO syndrome
RET	Medullary thyroid carcinoma/Multiple endocrine neoplasia IIA
RHBDF2	Tylosis with esophageal cancer
RNASEL	Prostate cancer 1
RUNX1	Leukemia, acute myeloid
SBDS	Shwachman-Diamond syndrome 1
SDHAF2	Paragangliomas 2
SDHB	Paragangliomas 4/gastrointestinal stromal tumor
SDHC	Paragangliomas 3 /Gastrointestinal tumor
SDHD	Paragangliomas 1,
SLX4	Fanconi anemia, complementation group P
SMAD4	Juvenile polyposis / hereditary hemorrhagic telangiectasia syndrome / Myhre syndrome
SMARCB1	Rhabdoid tumor
STK11	Peutz-Jeghers syndrome / pancreatic cancer / melanoma
SUFU	Medulloblastoma
TGFBR2	Loeys-Dietz syndrome 2 / esophageal cancer/colorectal cancer
TMEM127	Pheochromocytoma
TP53	colorectal cancer / glioma / osteosarcoma
TSC1	Tuberous sclerosis-1
TSC2	Tuberous sclerosis-2
VHL	Renal cell carcinoma
WRN	Werner syndrome
WT1	Wilms tumor
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C

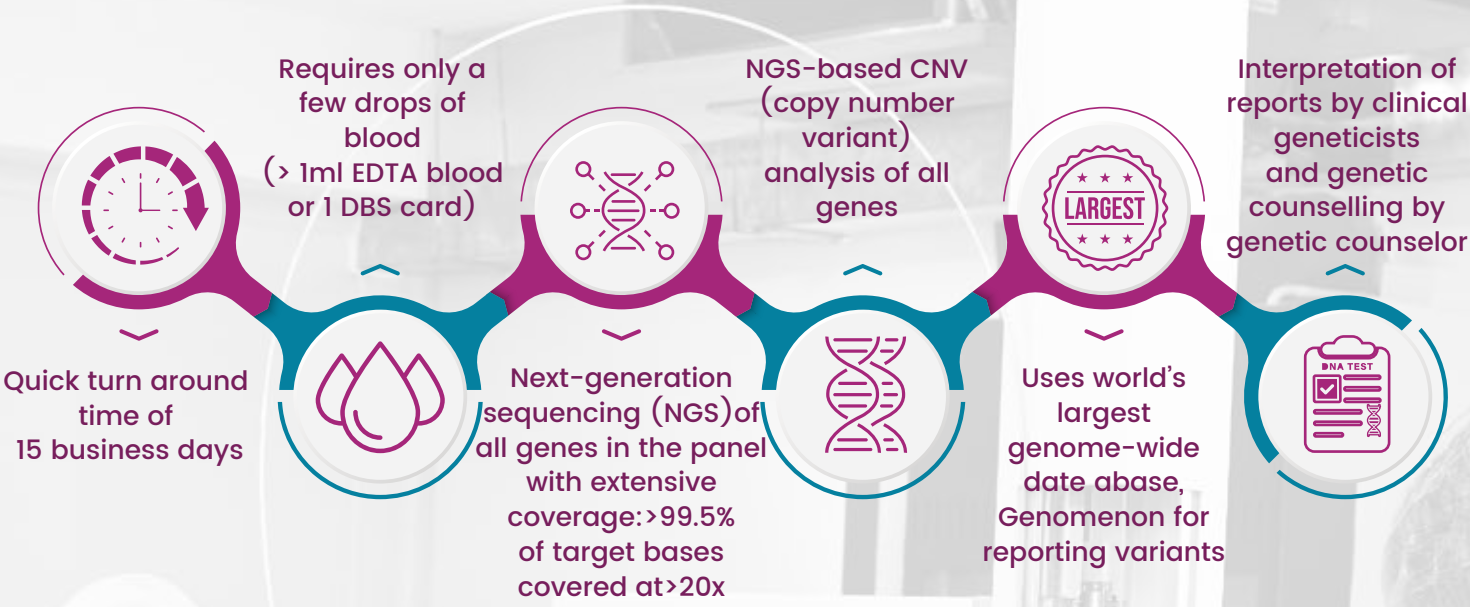
Who Should Consider This Panel ?

1. Individuals with breast cancer

2. Those with clinical or family history of a hereditary cancer syndrome like:

- Breast, ovarian, uterine, pancreatic, melanoma, sarcoma, and/or prostate cancer, particularly if early-onset (<50 years)
- Triple-negative breast cancer (<60 years)
- Male breast cancer
- Any known variant in a cancer susceptibility gene identified on tumour testing

What Makes LifeCell
A Leader In Genetic Testing



Test Ordering Information

Test Code	Test Name	Components	Specimen Type	Methodology	TAT
ONCO0065	OncoLife - Homologous Recombination Repair (HRR)	27 genes	3 ml peripheral blood in EDTA vial	NGS	21 Days
BCDMIHC	Breast Cancer Double Marker	ER,PR	Formalin fixed paraffin embedded tissue block/Biopsy	IHC	3-4 Days
BCM001	Breast Cancer Monitoring	CA15.3, CEA	2 ml serum in SST/Red Top (No Additive) tube	CLIA / CMIA	Same Day
BCQMIHC	Breast Cancer Quad	ER, PR, Her2neu, Ki-67	Formalin fixed paraffin embedded tissue block/Biopsy	IHC	4 Days
BCTMIHC	Breast Cancer Triple Marker	ER, PR, Her2neu	Formalin fixed paraffin embedded tissue block/Biopsy	IHC	3-4 Days
F0072	FISH for ERBB2 (HER2/neu) Amplification/Breast cancer & Gastric cancer	Her2neu Fish	Formalin fixed paraffin embedded tissue block/Biopsy	FISH	5 Days
B0059	BRCA1 & BRCA2 deletion/ duplication analysis	BRCA1, BRCA2	Formalin fixed paraffin embedded tissue block	MLPA	10 days
B0059a	BRCA1 & BRCA2 mutation analysis	BRCA1, BRCA2	3 ml EDTA vial-Peripheral blood/ FFPE Tissue/Paraffin block	NGS	20 days
GEN0487O	Hereditary cancer gene panel_ONCO	106 genes	Amniotic Fluid, Chorionic Villus Sample, Peripheral Venous Blood, Cord blood, Extracted DNA, Dry Blood Spot Whole blood- EDTA vacutainer DBS- DBS card AF -Sterile 15 ml Falcon tube CVS-Steril 15 ml Falcon tube with nutrient medium (provided by LifeCell) DNA-1.5ml sterile cryotube	NGS	21 days
P0055	PIK3CA mutation analysis (Exon 7, 9, 20)	PIK3CA	FFPE Tissue/Paraffin block Histopathology report must accompany the specimen	Sanger Sequencing	8 days
GEN0357O	BRCA1 & BRCA2 gene analysis	BRCA1 & BRCA2	"Peripheral Venous Blood, Extracted DNA - EDTA vacutainer Dry - DBS card Amniotic Fluid - Sterile 15 ml Falcon tube Corionic Villus Sample - Sterile 15 ml Falcon tube with nutrient medium (provided by LifeCell) DNA - 1.5ml sterile cryotube	NGS	21 Days
P0060	PDL1 -SP-142	PD-L1 SP142	Formalin fixed paraffin embedded tissue block/Biopsy	IHC	4 Days

Report Illustration

Test Name: WES hereditary cancer gene panel

Name:	Mrs. Dummy	Test Required	XXXXXXXX	Collected:	xx-xx-xxx	Status:	
Age:	XX Years	Case ID:	222XXXXX1233X	Received:	xx-xx-xxx 12:57	Sample Quality:	
Gender:	Female	Sample Type:	DEMO	Reported:	xx-xx-xxx 14:43	Client:	
Referring: Clinician	XXXXXXX						

CLINICAL INFORMATION/HISTORY

Proband presented with recurrent cancer of the right breast which was diagnosed in March 2020 as Invasive Ductal Carcinoma (IDC), Stage III, Triple Negative Breast Cancer (TNBC). She had a lumpectomy done in December 2019 and a modified radical mastectomy was done on the right breast in October 2020. IShe has 2 sons aged 18y and 14y. There is no significant family history on either side of your family.

RESULT SUMMARY

No pathogenic or likely pathogenic variants causative of the reported phenotype were identified
*Correlation with clinical profile and family history is required

FINDINGS RELATED TO PHENOTYPE

Gene & Transcript	Variant	Location	Zygosity	Disorder (OMIM)	Inheritance	Classification
BRCA1 NM_007294.4	c.4699G>A (p.Gly1567Arg)	Exon 15	Heterozygous	{Breast-ovarian cancer, familial, 1} (604370)	Autosomal Dominant	Uncertain Significance

VARIANT INTERPRETATIONS

BRCA1 chr17:41223232C>T - Uncertain Significance.

The missense variant NM_007294.4(BRCA1):c.4699G>A (p.Gly1567Arg) has been reported to ClinVar as Uncertain Significance with a status of (2 stars) criteria provided, multiple submitters, no conflicts (Variation ID 489723 as of 2022-03-03). The variant is observed in 2/30,616 (0.0065%) alleles from individuals of gnomAD South Asian background. The variant is observed in 1/5,008 (0.0200%) alleles from individuals of 1kG All background. There is a moderate physicochemical difference between glycine and arginine. The p.Gly1567Arg missense variant is predicted to be damaging by both SIFT and PolyPhen2. **For these reasons, this variant has been classified as Uncertain Significance.**
Germline pathogenic variants in the BRCA1 are associated with familial breast-ovarian cancer type 1 which is an autosomal dominant disorder. It is characterized with an increased risk for breast cancer (46%-87%), ovarian cancer (39%-63%), prostate cancer (9%) and pancreatic cancer (1%-3%). Breast cancer is one of the most common forms of cancer accounting for about 25% of all cancers in women. It is also more common in women than in men although men tend to have poorer outcomes due to delays in diagnosis. 5 to 10% of all breast cancers are inherited and most of them are associated with BRCA1 and BRCA2 genes.

RECOMMENDATIONS

The interpretation of this result should be done in the context of this individual's clinical and biochemical profile. Genetic counseling is recommended.

FINDINGS UNRELATED TO PHENOTYPE

Diagnostic findings not related to phenotype: No pathogenic variants in genes that are unrelated to the patient's phenotype were detected in this individual
Carrier status in genes related to disease: No pathogenic or likely pathogenic variants were detected. (Committee Opinion No. 691. American College of Obstetricians and Gynecologists. Obstet Gynecol 2017;129:e41-55)
Secondary findings: No pathogenic variants in genes related on the ACMG recommended secondary list were detected in this individual (Miller et al., 2021)

Gene/Transcript	Variant	Depth	Allelic depth	Alternate allele fraction	dbSNP rsID
BRCA1 (-)	c.4699G>A (p.Gly1567Arg)	96	53	44.79%	Not Available

Data quality statistics: Total data generated (Gb): 9, Panel coverage %: 99, Q30 bases: >85%

Mean Depth (X)	% of bases covered at		
99.03	1X	20X	30X
	97.92%	94.96%	93.17%

REFERENCES
1. Richards S. et al., Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, Genet Med., 17(5):405-24, 2015.
2. Freed D. et al., The Sentieon Genomics Tools-A fast and accurate solution to variant calling from next-generation sequence data. BioRxiv:115717, 2017.
3. https://cloud.google.com/life-sciences/docs/tutorials/deepvariant

Name of Salesperson: ..... Contact: .....