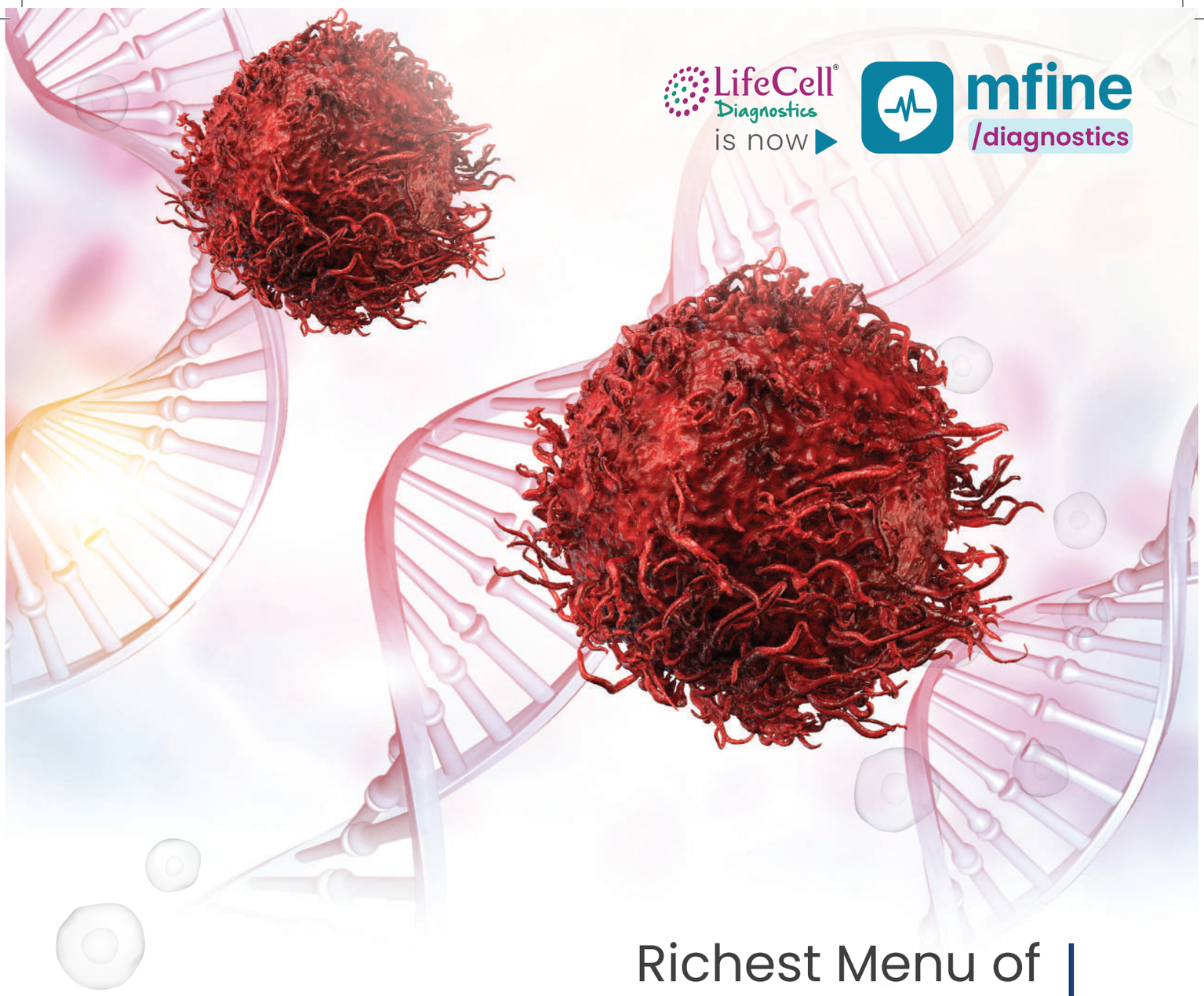


LifeCell® Diagnostics is now mfine /diagnostics

Richest Menu of

High-end specialized tests for

SOLID TUMORS

- Mammalife
- Pulmolife
- Cololife
- Gliomalife
- Oncolife Multigene
- Hematolife AML
- Hematolife MDS
- Hematolife CLL
- Hematolife CML
- Hematolife MPN
- Hematolife Multiple Myeloma
- Oncolife - Liquid Biopsy
- Pharmacolife
- Histolife

onco Life

Homologous Recombination Repair(HRR) 27 Genes Panel

Analyses mutations in HRR genes for potential therapeutic options and risks for other cancers

Clinical Utility

- Offers important value addition to the patient by identifying mutations in 27 genes which are predictive biomarkers for PARP inhibitor therapy.
- Next-Generation (NGS) on Illumina NovaSeq 6000 to identify germline mutations in a single assay
- Assay Detects: SNVs, Indels & Copy number variations
- Read depth: >250x

Highly Penetrant Genes:

BRCA1 | BRCA2 | CDK12 | FANCL | NBN | PPP2R2A | PTEN | RAD51 B | RAD51C | RAD51 D | RAD54L | TP53

Moderately Penetrant Genes:

ATM | BARD1 | BRIP1 | CHEK2 | FANCD21 | KRAS | MRE11 | PIK3CA | DOLD 1 | POLE | RAD50 | RAD5 | RAD52 | XRCC2

mamma Life HRR panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0068	Oncolife HRR 15 gene Germline panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDK12,CHEK1, CHEK2,FANCL, PALB2,PPP2R2A, RAD51B,RAD51C, RAD51D, RAD54L	NGS	21 Days	17000
ONCO0069	Oncolife HRR 15 gene Somatic panel	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDK12,CHEK1, CHEK2,FANCL, PALB2,PPP2R2A, RAD51B,RAD51C, RAD51D, RAD54L	NGS	21 Days	19000
ONCO0067	Oncolife HRR 15 Gene (Germline+Somatic)	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDK12,CHEK1, CHEK2,FANCL, PALB2,PPP2R2A, RAD51B,RAD51C, RAD51D, RAD54L	NGS	21 Days	31000
ONCO0065	OncoLife - Homologous Recombination Repair(HRR) 27 genes Panel	ATM,BARD1,BRCA1,BRCA2,BRIP1,CDK12,CHEK2,FANCD2,FANCL,KRAS,MRE11,NBN,PALB2,PIK3CA,POLD1,POLE,PPP2R2A,PTEN, RAD50,RAD51,RAD51B,RAD51C,RAD51D,RAD52, RAD54L,TP53, XRCC2	NGS	21 days	21000
ONCO0081	OncoLife - Targeted Breast Panel - Extended	SNVs/Indels, CNA - ATM, BARD1, BRCA1, BRCA2, BRIP1, CDK12, CHEK1, CHEK2, ERBB2, FANCL, PIK3CA, PPP2R2A, RAD51C, RAD45L	NGS	21 Days	20000
P0060	PDL1 -SP-142	PD-L1 SP142	IHC	4 Days	8500

Report Illustration

Oncolife HRR (15 Genes)

PERSONAL/ FAMILY HISTORY

	Proband	Immediate Relatives (Parents, Siblings)	Son/Daughter	Paternal/Maternal Relatives
Relationship	Self	Mother	-	-
Cancer Type	Invasive carcinoma of Breast	Ovarian Cancer	-	-
Age at diagnosis (in years)	32	46	-	-

Family history as provided by qualified healthcare provider on the test request form:

GENOMIC HIGHLIGHT

POSITIVE

Pathogenic or disease-causing variant was identified

Description of variants classifications based on Published Literature and ACMG Guidelines

POSITIVE

Identified variant is known to elevate the cancer risk.

Already known as Pathogenic variant

Please consult with your doctor to create a screening and management plan and to identify relatives who may need to be tested.

YUS

Identified variant is presently not known to increase the cancer risk.

The genetic/clinical data is insufficient for us to analyze the mutation or to associate it with any kind of a clinical condition.

Patient may or may not develop any associated condition, and its risk cannot be predicted due to inadequate scientific reported evidence.

NEGATIVE

Ito clinically relevant variant was identified that is associated with elevated cancer ask.

Please consult with your doctor to discuss the surveillance recommendations.

STATUS OF HRR GENES

Below is a table enlisting HRR genes and the status of mutations in this patient:

Gene Name	Detected Mutation
ATM	Not detected
BARD1	Not detected
BRCA1	Not detected
BRCA2	Pathogenic

Gene Name	Detected Mutation
BRIP1	Not detected
CDK12	Not detected
CHEK1	Not detected
CHEK2	Not detected

Gene Name	Detected Mutation
FANCL	Not detected
PALB2	Not detected
PPP2R2A	Not detected
RAD51B	Not detected

Gene Name	Detected Mutation
RAD51C	Not detected
RAD51D	Not detected
RAD54L	Not detected

For all the HRR genes analyzed in the above-mentioned table, variants were classified as:

- 'Pathogenic/ Likely pathogenic' - variants for the gene were deleterious or disease-causing
- 'VUS' - variants for the gene were of uncertain significance
- 'Not detected' - variants for the gene were not detected in the patient

Note: This test only mentions the status of the HRR genes in our panel, The results mentioned are not an equivalent of the HRD status and/or positive Genomic Instability Score.

mamma Life

For diagnosis & therapy management of
Hereditary & Sporadic Breast Cancer

ONCO0071 | Oncolife Comprehensive Hereditary Cancer gene panel +MLPA

Pipelines are capable enough to pick up SNVs, INDELs, CNVs

Comprehensive panels provide a genetic testing of Hereditary Cancers

Digital MLPA kits for 25 genes which can be used to identify any deletions or duplications

Identifies patients most likely to benefit from poly ADP - Ribose Polymerase (PARP) inhibitors

mamma Life Hereditary panels

Test code	Test Name	Components	Technique	TAT	MRP
B0059	BRCA1 & BRCA2 deletion/duplication analysis	BRCA1, BRCA2	MLPA	10 days	15000
B0059a	BRCA1 & BRCA2 mutation analysis	BRCA1, BRCA2	NGS	20 days	20000
GEN0357O	BRCA1 & BRCA2 gene analysis	BRCA1 & BRCA2	NGS	21 Days	13000
P0055	PIK3CA mutation analysis (Exon 7, 9 , 20)	PIK3CA	Sanger Sequencing	8 days	7000
GEN0487_ONCO	Hereditary cancer gene panel_ONCO	AIP, ALK, APC, AR, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CD82, CDC73, CDH1, CDK4, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, EGFR, ELAC2, ENG, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, HRAS, KIT, MAX, MEN1, MET, MLH1, MLH3, MRE11A, MSH2, MSH3, MSH6, MSR1, MUTYH, MXI1, NBN, NF1, NF2, NSD1, PALB2, PHOX2B, PMS1, PMS2, PRF1, PRKAR1A, PTCH1, PTEN, RAD5, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RNASEL, RUNX1, SBDS, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TGFBR2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC	NGS	21 days	21000
ONCO0071	Oncolife comprehensive Hereditary Cancer gene panel +MLPA	Hereditary cancer gene panel_ONCO (WGS) + Deletion Duplication analysis Digital MLPA (25 genes)	NGS +MLPA	21 days	27000

Report Illustration

WES Hereditary cancer gene panel - (106 genes)

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

Gene	Percentage of Coding Region Covered	Gene	Percentage of Coding Region Covered	Gene	Percentage of Coding Region Covered
AIP	100	ALK	100	APC	99.02
AR	100 A	TM	94.58	BAP1	100
BARD1	98.94	BLM 9	4.08	BMPR1A	100
BRCA1	97.95	BRCA2	97.37	BRIP1	99.98
BUB1B	100	CD82	100	CDC73	100
CDH1	99.16	CDK4	100	CDKN1C	100
CDKN2A	100	CEBPA	100	CEP57	98.82
CHEK2	90.91	CYLD	94.02	DDB2	100
DICER1	100	DIS3L2	98.16	EGFR	99.86
ELAC2	100	ENG	100	EPCAM	81.48
ERCC2	100	ERCC3	100	ERCC4	99.9
EXT1	100	EXT2	98.37	EZH2	95.06
FANCA	97.75	FANCB	95	FANCC	90.41
FANCD2	94.91	FANCE	100	FANCF	100
FANCG	100	FANCI	100	FANCL	84.83
FANCM	86.76	FH	100	FLCN	100
GATA2	100	GPC3	100	HRAS	100
KIT	100	MAX	100	MEN1	100
MET	100	MLH1	98.9	MLH3	100
MSH2	96.15	MSH3	95.61	MSH6	100
MSR1	100	MUTYH	100	MXI1	100
NBN	95.59	NF1	97.64	NF2	100
NSD1	100	PALB2	100	PHOX2B	100
PMS1	90.79	PMS2	100	PRF1	100
PRKAR1A	95.9	PTCH1	100	PTEN	89.95
RAD51C	100	RAD51D	100	RB1	80.13
RECQL4	100	RET	100	RHBDF2	100
RNASEL	100	RUNX1	97.48	SBDS	00
SDHAF2	98.98	SDHB	100	SDHC	100
SDHD	80.27	SLX4	100	SMAD4	98.19
SMARCB1	100	STK11	95.45	SUFU	100
TGFBR2	100	TMEM127	100	TP53	100
TSC1	100	TSC2	100	VHL	100
WRN	88.7	WT1	100	XPA	97
XPC	100				

pulmo Life

For diagnosis & therapy management of **Lung Cancer**

Patient Selection- Diagnosed with Lung Cancer

Technology- NGS (Novaseq & Sanger)

Specimen- Formalin fixed paraffin embedded tissue block

Clinical Utility

- Provides a comprehensive genetic profile of Lung cancer & its subtypes.
- Identifies mutations that help in taking therapy related decisions according to the latest WHO guidelines
- Prediction of patient's sensitivity to checkpoint inhibitor immunotherapies

pulmo Life panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0048	OncoLife - Targeted Lung panel - Basic (Sanger)	EGFR, ALK, ROS1	Sanger sequencing, FISH	10 days	13000
ONCO0066	OncoLife Targeted Lung Panel Extended with PDL1 DAKO	Hotspot genes: ALK, BRAF, EGFR, ERBB2, KRAS, MAP2K1, MET, NRAS, PIK3CA, ROS1, NTRK, TP53 Fusion genes: ALK, RET, ROS1, MET exon 14 skipping IHC: PDL1 22c3 Dako	NGS, IHC	14 Days	28000
ONCO0049	OncoLife Targeted Lung Panel Extended with PDL1 SP263	Hotspot genes: ALK, BRAF, EGFR, ERBB2, KRAS, MAP2K1, MET, NRAS, PIK3CA, ROS1, NTRK, TP53 Fusion genes: ALK, RET, ROS1, MET exon 14 skipping IHC: PDL1 SP263 Ventana	NGS, IHC	14 Days	25000
ONCO0082	OncoLife - Targeted Lung Panel - Extended without PD-L1	SNVs/Indels - ALK, BRAF, CDKN2A, EGFR, ERBB2, KRAS, MAP2K1, MET, NRAS, PIK3CA, RET, ROS1, STK11 Fusions - ALK, ROS1, RET, MET exon 14 skipping mutation CNA -CDKN2A, EGFR, ERBB2, STK11	NGS	21 Days	15000
ONCO0061	OncoLife - Focus gene panel	52 genes	NGS	4 weeks	44000
ONCO0094	OncoLife - Hotspot 50 gene panel	SNV/Indels: ABL1,AKT1,ALK, APC, ATM,BRAF,CDH1,CDKN2A, CSF1R,CTNNB1,EGFR,ERBB2,ERBB4,EZH2,FBXW7, FGFR1,FGFR2,FGFR3,FLT3,GNA11,GNAQ,GNAS,H NF1A,HRAS,IDH1,IDH2,JAK2,JAK3,KDR,KIT,KRAS, MET,MLH1,MPL,NOTCH1,NPM1,NRAS,PDGFRA,PIK 3CA,PTEN,PTPN11,RB1,RET,SMAD4,SMARCB1, SMO, SRC, STK11, TP53,VHL	NGS	15 Days	24000

Report Illustration

OncoLife - Targeted Lung Panel Extended

VARIANT DETAILS

Gene	Variant	Variant Details	Clinical significance	Variant Type	Reference
EGFR	chr7 g 55242467 55242485del insT, ENST00000275493, Exon 19	c2237 2255delist, p.G1u746 5er752, 20%	Pathogenic	Inframe deletion	ACMG guidelines

GENOMICS FINDING FROM TUMOR PROFILING

Genomic Alteration

EGFR Exon 19 (p.Glu746_Ser752delinsVal Allelic burden 20%

Relevant Therapies (In same cancer type)

Osimertinib	**Relevance** Responsive
Erlotinib	Responsive
Gefitinib	Responsive
Afatinib	Responsive
Dacomitinib	Responsive
	Responsive

Relevant Therapies (In different cancer)

y NA **Relevance** NA **Type** NA

• *NA: Not Available*

STATUS OF MUTATIONS IN CANCER RELATED BIOMARKERS

GENE	BRAF	MET	KRAS	EGFR	ERBB2	ALK	ROS1	CDKN2A	
STATUS	Not detected	Not detected	Not detected	Pathogenic	Not detected	Not detected	Not detected	VUS	
GENE	**STK11**	**RET**							
STATUS	Not detected	Not detected							

GENES EVALUATED

Onco Life Comprehensive Targeted Lung panel detects mutations (SNVs and Short Indels), Copy Number Variations (CNVs), gene fusions and splice variants in the 15 genes :

GENE LIST

SNVs/InDels Covered in **OnceLife**

ALK | BRAF | CDKN2A | EGFR | EPCAM | ERBB2 | KRAS | MET | MLH1 | MSH2 | MSH6 | PMS2 | RET | ROS1 | STK11

CNAs/InDels Covered in **OnceLife**

CDKN2A | EGFR | EPCAM | ERBB2 | MET | MLH1 | MSH2 | MSH6 | PMS2 | STK11

Gene Covered in **OnceLife**

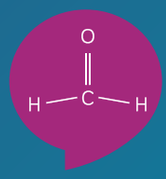
ALK | MET | RET | ROS1

colo Life

For diagnosis & therapy management of **Colon Cancer**

Patient Selection- Patients with colorectal cancer or patients shortlisted for immunotherapy

Technology- NGS (Novaseq, Sanger, IHC)



Specimen- Formalin fixed paraffin embedded tissue block

Clinical Utility

- Identification of sporadic and hereditary colorectal cancers (HNPCCs) and Lynch Syndrome
- Gold standard technique for the detection of mutations in mismatch repair (MMR) genes
- Prediction of patient's sensitivity to immune checkpoint inhibitors

colo Life panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0034	OncoLife - Targeted gene panel - Basic- Colorectal (NGS)	KRAS, NRAS, BRAF	NGS	14 days	10000
ONCO0035	OncoLife - Targeted Colorectal panel - Extended	KRAS, NRAS, BRAF, MSI	NGS, Fragment analysis	14 days	14500
ONCO0036	OncoLife - Targeted Colorectal panel - Extended (NGS)	KRAS, NRAS, BRAF, MMR-IHC	NGS, IHC	14 Days	12000
M0039a	Microsatellite instability (MSI) testing	MLH1, MSH2, MSH6 and PMS2	Fragment analysis	10 days	9000
GEN0359_ONCO	Lynch Syndrome/ HNPCC gene panel_ONCO	MLH1, MSH2, MSH6, PMS1, EPCAM, MSH3, MLH3, PMS2	NGS	21 days	13000
ONCO0009	Microsatellite Instability Panel for Colorectal Carcinoma by IHC	MSH2, MSH6, MLH1 and PMS2	IHC - Manual	7 days	7000
ONCO0092	OncoLife - Colocomprehensive panel	APC, AKT1, BRAF, ERBB2, EZH2, GNAS, HRAS, KRAS, NRAS, PIK3CA, PTEN, SMAD4, SMO, STK11, TP53 + MSI (IHC)	NGS,IHC	15 days, 7 days (IHC)	16500
ONCO0093	TP53 hotspot mutation analysis	TP53	NGS	15 days	10000

Report Illustration

OncoLife - Colo Comprehensive Panel

SUMMARY OF FINDINGS

Variant	Gene	AA	Effect	Cvg (%Alt)	COSMIC	ClinVar	AMP/ASCO/CAP
chr12:25398281(C>T)	KRAS[1]	G13D	Missense	417 (19.9%)	COSM532	Pathogenic	IA

VARIANT CLASSIFICATION

We continually perform ongoing evaluations of variant classifications. When new evidence about a variant is identified and determined to result in clinical significance and management change, that information will automatically be made available to the healthcare provider through an amended report.

Methods and Limitations

METHODOLOGY

The individual's DNA was extracted and fragmented, for the coding regions of the selected panel (AmpliSeq for Illumina Cancer Hotspot Panel v2) which was further targeted for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the GENEYX analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

VARIANT ASSESSMENT PROCESS

The following databases and in-silico algorithms are used to annotate and evaluate the impact of the variant in the c text of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, SIFT, PolyPhen2, GERP, LRT PRED, Civic. Analysis was reported using the to HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches that used most frequently by the clinical labs submitting to ClinVar.

GENE LIST (SNVS/INDELS)

ABL1 | AKT1 | ALK | APC | ATM | BRAF | CDH1 | CDKN2A | CSF1R | CTNNB1 | EGFR | ERBB2 | ERBB4 | EZH2 FBXW7 | FGFR1 | FGFR2 | FGFR3 | FLT3 | GNA11 | GNAQ | GNAS | HNF1A | HRAS | IDH1 | IDH2 | JAK2 | JAK3 | KDR KIT | KRAS | MET | MLH1 | MPL | NOTCH1 | NPM1 | NRAS | PDGFRA | PIK3CA | PTEN | PTPN11 | RB1 | RET | SMAD4 SMARCB1 | SMO | SRC | STK11 | TP53 | VHL

glioma Life

For diagnosis & therapy management of
CNS Tumors

Patient Selection- Patients with Adult & Pediatric Central Nervous System (CNS) tumors

Technology- NGS (Novaseq, Sanger)

Specimen- Formalin fixed paraffin embedded tissue block

Clinical Utility

- Provides a comprehensive genetic profile of CNS tumor subtypes such as glioma, medulloblastoma, meningioma and ependymoma.
- Identifies mutations that help differentiate between low and high grade gliomas according to the latest WHO guidelines.

glioma Life panels

Test code	Test Name	Components	Technique	TAT	MRP
M0038	MGMT promoter methylation analysis	MGMT	Methylation PCR	16 days	9000
I0024c	IDH1/2 mutation analysis	IDH1/IDH2(exon 4)	NGS	14 days	9000
ONCO0062	OncoLife - Targeted Melanoma panel - Basic	KIT, BRAF , NRAS	NGS	14 days	10000
ONCO0064	OncoLife - Glioma panel extended	IDH1/IDH2 (exon 4), TP53 (IHC), ATRX (IHC),EGFR amplification	NGS, IHC, FISH	14 days	50000
ONCO0038	OncoLife - Theranostic Glioma panel	IDH1/IDH2 (exon 4), codeletion 1p/19q, MGMT (3 genes)	NGS, FISH, Methylation PCR	14 days	26000
ONCO0009	Microsatellite Instability Panel for Colorectal Carcinoma by IHC	MSH2, MSH6, MLH1 and PMS2	IHC - Manual	7 days	7000

Report Illustration

OncoLife - Glioma Panel Extended

SUMMARY OF FINDINGS

Variant	Gene	AA	Effect	Cvg (%Alt)	COSMIC	ClinVar	AMP/ASCO/CAP
chr15:90631839(T>C)	IDH2[1]	R172G	Missense	198(11.11%)	COSM33733	Likely Pathogenic	IA

MAIN FINDINGS: SIGNIFICANT VARIANT (S) IDENTIFIED IN IDH2

Clinical significance of IDH2 in Cancer: IDH2 mutations have been observed in a number of cancer types, including sarcomas, hematologic malignancies, colon cancer and brain cancer. Mutations in the two isocitrate dehydrogenase enzymes involved in cytoplasmic (IDH1) and mitochondrial (IDH2) conversion of alpha-ketoglutarate to D-2-hydroxyglutarate have been described as mutually exclusive in many of these cancer types. The most frequent mutations involve R132 (IDH1) and R172 (IDH2) involve the active site and result in neomorphic enzyme activity. Although IDH2 (R172) mutations are associated with poorer overall prognosis in AML patients, its utility as a prognostic marker in MDS is still under debate. Additionally, IDH2 (R140) has been associated with improved overall survival in AML. IDH2 mutations have been associated with improved prognosis in gliomas.

CIVIC VARIANT SUMMARY

IDH2 R172 is a gain-of-function mutation commonly observed in acute myeloid leukemia and is enriched in those patients which are cytogenetically normal. R172 is analogous to IDH1 R132 mutations and leads to neomorphic enzymatic activity. Specifically this mutation" catalyzes the conversion of alpha ketoglurarate to betahydrozyglutarate" leading to increased levels of beta-hydrozyglutarate. This in turn leads to hypermethylation of target genes and an inhibition of cellular differentiation. R172 mutations generally lead to worse overall survival and higher rates of relapse compared to the most common IDH2 mutations in AML (R140).

VARIANT CLASSIFICATION

We continually perform ongoing evaluations of variant classifications. When new evidence about a variant is identified and determined to result in clinical significance and management change, that information will automatically be made available to the healthcare provider through an amended report.

Methods and Limitations

METHODOLOGY

The individual's DNA was extracted and fragmented, with fragments from the coding regions from an inhouse developed targeted gene panel for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the GENEYX analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

VARIANT ASSESSMENT PROCESS

The following databases and in-silico algorithms are used to annotate and evaluate the impact of the variant in the context of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, SIFT, PolyPhen2, GERP, LRT PRED, Civic. Analysis was reported using the to HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches that used most frequently by the clinical labs submitting to ClinVar.

GENES EVALUATED: IDH1, IDH2

IDH1 | IDH2

gastri Life

For diagnosis & therapy management of **Gastric & Intestinal Cancers**

Patient Selection-Patients with Adult & Pediatric Central Nervous System (CNS) tumors

Technology- NGS (Novaseq, Sanger)

Specimen - Formalin fixed paraffin embedded tissue block

Clinical Utility

- Comprehensive panel for diagnosis and identifying targeted therapies for patients with gastrointestinal stromal tumors
- Detects activating mutations in KIT and PDGFRA.
- Assessment of microsatellite instability

gastri Life panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0006	GIST Panel by IHC	IHC	IHC - Manual	7 days	7000
K0014a	KIT mutation analysis (Exon 9, 11, 13, 17)	KIT	Sanger Sequencing	14 days	4000
ONCO0028	OncoLife - Tageted gene panel - GIST	*CD117 *DOG-1 *ERBB2 *KIT *BRAF *PDGFRA *TP53	IHC, NGS	21 days	16000
ONCO0029	OncoLife - Targeted Lung panel - Basic	KIT & PDGFRA (2 genes)	NGS	14 days	20000
ONCO0032	OncoLife - Theranostic gene panel -GIST (Gastrointestinal Stromal Tumor)	KIT & PDGFRA	Sanger Sequencing	14 days	14000

Report Illustration

OncoLife - Theranostic GIST Panel

SUMMARY OF FINDINGS

Variant	Gene	AA	Effect	Cvg (%Alt)	COSMIC	ClinVar	AMP/ASCO/CAP
chr17:7578205(C>T)	TP531	S215N	Missense	118(33.9%)	COSM44093	Likely Pathogenic	IA

MAIN FINDINGS: SIGNIFICANT VARIANT(S) IDENTIFIED IN TP53

Clinical significance of TP53 in Cancer: TP53 mutations are universal across cancer types. The loss of a tumor suppressor ismost often through large deleterious events, such as frameshift mutations, or premature stop codons. In TP53 however, many of the observed mutations in cancer are found to be single nucleotide missense variants. These variants are broadly distributed throughout the gene, but with the majority localizing in the DNA binding domain. There is no single hotspot in the DNA binding domain, but a majority of mutations occur in amino acid positions 175, 245, 248, 273, and 282 (NM_000546) (Olivier et al., 2010). To fulfill its proper biological function four TP53 polypeptides must form a tetramer which functions as a transcription factor, therefore even if one out of four polypeptides has inactivating mutation it may lead to dominant negative phenotype of variable degree. While a large proportion of cancer genomics research is focused on somatic variants, TP53 is also of note in the germline. Germline TP53 mutations are the hallmark of Li-Fraumeni syndrome, and many (both germline and somatic) variants have been found to have a prognostic impact on patient outcomes. The significance of many polymorphisms for susceptibility and prognosis of disease is still very much up for debate.

VARIANT CLASSIFICATION

We continually perform ongoing evaluations of variant classifications. When new evidence about a variant is identified and determined to result in clinical significance and management change, that information will automatically be made available to the healthcare provider through an amended report.

Methods and Limitations

METHODOLOGY

The individual's DNA was extracted and fragmented, for the coding regions of the selected panel (AmpliSeq for Illumina Cancer Hotspot Panel v2) which was further targeted for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the GENEYX analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

GENE LIST (SNVS/INDELS)

ABL1 | AKT1 | ALK | APC | ATM | BRAF | CDH1 | CDKN2A | CSF1R | CTNNB1 | EGFR | ERBB2 | ERBB4 | EZH2 | FBXW7 FGFR1 | FGFR2 | FGFR3 | FLT3 | GNA11 | GNAQ | GNAS | HNF1A | HRAS | IDH1 | IDH2 | JAK2 | JAK3 | KDR | KIT KRAS | MET | MLH1 | MPL | NOTCH1 | NPM1 | NRAS | PDGFRA | PIK3CA | PTEN | PTPN11 | RB1 | RET SMAD4 | SMARCB1 SMO | SRC | STK11 | TP53 | VHL

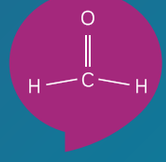
onco Life

TruSight Oncology 500 (illumina) **Gene Panels**

- **100% coverage** of ESMO guidelines for 9 tumor types
- **100% coverage** of NCCN guidelines for 11 tumor types
- Covers biomarkers for **47 drug labels**
- **95%** analytical sensitivity
- **99.9%** analytical specificity for small variants

Patient with metastatic cancers & unknown primaries

Technology- NGS (Novaseq)



Specimen- Formalin fixed paraffin embedded tissue block

Clinical Utility

- 523 cancer relevant genes, 55 fusions, TMB & MSI
- The test has very high depth of coverage, low TAT and a detailed report matching patients to FDA approved drugs and clinical trials makes this test of high utility to patients.
- TSO 500 combine comprehensive genomic content with sophisticated informatics algorithm to provide accurate TMB estimation that is highly Concordant with whole-exome studies

onco Life TSO panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0073	OncoLife - TSO 500 gene panel	523 genes for DNA variants, 55 genes for RNA fusions and splice variants, TMB, MSI	NGS	21 days	149000
ONCO0076	OncoLife - TSO 500 gene panel + PDL1	523 genes for DNA variants, 55 genes for RNA fusions and splice variants, TMB, MSI, PDL1 by IHC	NGS,IHC	21 days	159000
ONCO0055	OncoLife - Comprehensive gene panel DNA variants+ Tumor mutation burden [TMB] + Microsatellite Instability (MSI)	523 genes for DNA variants, TMB, MSI, PDL1 by IHC	NGS,IHC	21 days	75000
ONCO0060	OncoLife - Comprehensive Plus gene panel + Tumor mutation burden [TMB] + Microsatellite Instability (MSI)	553 genes +TMB+ MSI	NGS	4 weeks	140000
ONCO0063	Oncolife Solid Tumor RNA Fusion panel	299 gene associated with gene fusions	NGS	14 Days	40000

Report Illustration

OncoLife - Tru Sight Oncology 500 (illumina) Gene Panel

SUMMARY OF GENOMIC AND BIOMARKER FINDINGS

Detected biomarkers with therapy implications

BIOMARKER	VAF (%)	APPROVED TREATMENTS FORPATIENT DISEASE	BIOMARKER SCORE	TRIALS	OTHER TREATMENTS	DRUG APPROVAL	BIOMARKER SCORE	TRIALS
TMB-L 9.40 mut/Mb	-	No therapies or clinical trials related to this biomarker						
MSI-L	-	No therapies or clinical trials related to this biomarker						
Additional biomarkers#		Diagnostic: SSBP2/EWSR1 (IB, 6)						

ADDITIONAL TEST RESULTS

List of biomarkers identified from additional tests

BIOMARKER	ASSAY	COMMENT
MSI-L 3.17 (MSI)	DNA sequencing	MSI-Low
TMB-L 9.40 mut/Mb (TMB)	DNA sequencing	TMB-Low

VARIANT	CODING	DNA TYPE AND EFFECT	VAF (%)	CLASSIFICATION
BRCA2 p.T3085fs	ENST00000380152.3 c.9253dup	ins Frameshift	9.00	Pathogenic
DICER1 p.Q1774*	ENST00000343455.3 c.5320C>T	SNV Nonsense	7.89	Pathogenic
HNF1A p.P291fs	ENST00000257555.6 c.864del	del Frameshift	25.42	Pathogenic
MITF	ENST00000448226.2 c.881-1G>A	SNV Splice site	23.81	Pathogenic
AMER1 p.F173fs	ENST00000330258.3 c.519del	del Frameshift	25.95	Likely pathogenic
AR p.E128fs	ENST00000374690.3 c.381del	del Frameshift	14.93	Likely pathogenic
ARID1A p.Q546fs	ENST00000324856.7 c.1636del	del Frameshift	24.80	Likely pathogenic
ATM p.K1904fs	ENST00000278616.4 c.5712del	del Frameshift	29.50	Likely pathogenic
BCORL1 p.N1387fs	ENST00000218147.7 c.4160dup	ins Frameshift	32.52	Likely pathogenic
BCORL1 p.P206fs	ENST00000218147.7 c.617del	del Frameshift	26.99	Likely pathogenic
FANCD2 p.I1155fs	ENST00000383807.1 c.3463del	del Frameshift	28.93	Likely pathogenic
IFNGR1 p.S378fs	ENST00000367739.4 c.1132_1133del	del Frameshift	47.06	Likely pathogenic
MED12 p.H1729fs	ENST00000374080.3 c.5186_5187del	del Frameshift	7.87	Likely pathogenic
MRE11 p.N511fs	ENST00000323929.3 c.1532del	del Frameshift	27.74	Likely pathogenic
NSD1 p.M1531fs	ENST00000439151.2 c.4591del	del Frameshift	8.82	Likely pathogenic
RNF43 p.G659fs	ENST00000407977.2 c.1976del	del Frameshift	37.31	Likely pathogenic
RNF43 p.V490fs	ENST00000407977.2 c.1468del	del Frameshift	31.64	Likely pathogenic
SETD2 p.S1350fs	ENST00000409792.3 c.4048del	del Frameshift	26.19	Likely pathogenic

GENE COVERAGE- 523 DNA MUTATIONS + 55 FUSIONS + TMB+MSI

onco Life /multigene

Multigene assay including all relevant mutations (SNVs, indels, CNVs & Fusions) for possible therapeutic associations as per FDA approved therapies for the reported alterations

Patients with aggressive cancers having limited treatment options & refractory patients

Technology- NGS (Novaseq)

Specimen- Formalin fixed paraffin embedded tissue block

onco Life /multigene panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0057	OncoLife - Hotspot 50 gene panel	**SNV/Indels**: ABL1, AKT1, ALK, APC, ATM ,BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7,F GFR1, FGFR2, FGFR3, FLT3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53,VHL	NGS	15days	24000
ONCO0075	OncoLife - Focus gene panel	52 genes	NGS	4 weeks	44000
ONCO0057	OncoLife - Comprehensive gene panel + Tumor mutation burden [TMB] + Microsatellite Instability (MSI) + PDL1	212 genes +TMB + MSI+PD-L1	NGS + Sanger Sequencing	4 weeks	100000
ONCO0063	Oncolife Solid Tumor RNA Fusion panel	299 gene associated with gene fusions	NGS	14 Days	40000
ONCO0077	OncoLife - Targeted 1212 gene panel	1064 genes for SNVs/Indels, 570 genes for Copy Number Alterations, 299 genes associated with Gene-Fusions, TMB, MSI, PDL1 by IHC	NGS, IHC	21 days	95000
ONCO0078	OncoLife - Targeted Liquid biopsy 1212 gene panel	1064 genes for SNVs/Indels, 570 genes for Copy Number Alterations, 299 genes associated with Gene-Fusions, TMB, MSI, PDL1 by IHC	NGS,IHC	21 days	95000

Report Illustration

OncoLife - Comprehensive Plus Gene Panel + Tumor Mutation Burden [TMB] + Microsatellite Instability (MSI)

SUMMARY OF FINDINGS

Variant	Gene	AA	Effect	Cvg (%Alt)	COSMIC	ClinVar	AMP/ASCO/CAP
chr12:25398281(C>T)	KRAS1	G13D	Missense	417 (19.9%)	COSM532	Pathogenic	IA

MAIN FINDINGS: SIGNIFICANT VARIANT(S) IDENTIFIED IN KRAS

Clinical significance of KRAS in Cancer: Mutations in the RAS family of proteins are frequently observed across cancer types. The amino acid positions that account for the overwhelming majority of these mutations are G12, G13 and Q61. The different protein isoforms, despite their raw similarity, also behave very differently when expressed in non-native tissue types, likely due to differences in the C-terminal hyper-variable regions. Mis-regulation of isoform expression has been shown to be a driving event in cancer, as well as missense mutations at the three hotspots previously mentioned. While highly recurrent in cancer, attempts to target these RAS mutants with inhibitors have not been successful, and has not yet become common practice in the clinic. The prognostic implications for KRAS mutations vary between cancer types, but have been shown to be associated with poor outcome in colorectal cancer, non-small cell lung cancer, and others.

There are no FDA-approved or NCCN-compendium listed treatments specifically for patients with colon cancer harboring activating RAS mutation(s). However, laboratory and preliminary clinical data suggest that KRAS-mutant cancers may be sensitive to MEK- or ERK-targeted inhibitors [PMID: 20921465, PMID: 21228335, PMID: 20619739, PMID: 24024839, PMID: 18316791]. Patients with any known KRAS mutation (exon 2, 3, 4) or NRAS mutation (exon 2, 3, 4) should not be treated with either Cetuximab or Panitumumab [NCCN Guidelines Version 1.2022 Colon Cancer page no 22].

VARIANT CLASSIFICATION

We continually perform ongoing evaluations of variant classifications. When new evidence about a variant is identified and determined to result in clinical significance and management change, that information will automatically be made available to the healthcare provider through an amended report.

Methods and Limitations
METHODOLOGY

The individual's DNA was extracted and fragmented, for the coding regions of the selected panel (AmpliSeq for Illumina Cancer Hotspot Panel v2) which was further targeted for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the GENEYX analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

VARIANT ASSESSMENT PROCESS

The following databases and in-silico algorithms are used to annotate and evaluate the impact of the variant in the context of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, SIFT, PolyPhen2, GERP, LRT PRED, Civic. Analysis was reported using the to HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches that used most frequently by the clinical labs submitting to ClinVar.

GENE LIST (SNVS/INDELS)

ABL1 | AKT1 | ALK | APC | ATM | BRAF | CDH1 | CDKN2A | CSF1R | CTNNB1 | EGFR | ERBB2 | ERBB4 | EZH2 | FBXW7 | FGFR1 | FGFR2 | FGFR3 | FLT3 | GNA11 | GNAQ | GNAS | HNF1A | HRAS | IDH1 | IDH2 | JAK2 | JAK3 | KDR | KIT | KRAS | MET | MLH1 | MPL | NOTCH1 | NPM1 | NRAS | PDGFRA | PIK3CA | PTEN | PTPN11 | RB1 | RET | SMAD4 | SMARCB1 | SMO | SRC | STK11 | TP53 | VHL

GENES EVALUATED

APC | AKT1 | BRAF | ERBB2 | EZH2 | GNAS | HRAS | KRAS | NRAS | PIK3CA | PTEN | SMAD4 | SMO | STK11 | TP53

Onco Life Liquid Bx.

Highly sensitive liquid biopsy assays utilizing both digital PCR (dPCR) and targeted Next-generation Sequencing (NGS) to enable cancer driver identification, serial monitoring, and recurrence detection.

Analyze cell-free nucleic acid (cfNA)

from cell-free DNA and RNA to identify primary cancer driver and resistance mutations.

Patients with metastatic solid-tumors & insufficient tissue

Technology- Droplet digital PCR & Novaseq (NGS)

Specimen- 10ml whole blood in streck tube

onco Life Liquid Bx. panels

Test code	Test Name	Components	Technique	TAT	MRP
ONCO0053	OncoLife - Liquid Bx. Cell Free RAS hotspot mutation analysis	NRAS, KRAS	Droplet digital PCR	4 weeks	19000
ONCO0050	OncoLife - Liquid Bx. Cell Free EGFR hotspot mutation analysis	EGFR (T790M, L858R, Exon 19 deletion)	Droplet digital PCR	4 weeks	11000
ONCO0051	OncoLife - Liquid Bx. Cell Free BRAF hotspot mutation analysis	BRAF	Droplet digital PCR	4 weeks	16000
ONCO0052	OncoLife - Liquid Bx. Cell Free KRAS hotspot mutation analysis	KRAS	Droplet digital PCR	4 weeks	4000
ONCO0054	OncoLife - Liquid Bx. Cell Free Lung cancer panel	12 genes	NGS	4 weeks	35000
ONCO0039	OncoLife - Liquid biopsy 52 gene panel	52 genes	NGS	4 weeks	55000

onco Life Exomes

Multigene exome analysis to identify variants inherited from the parents causing recessive or dominant disease.

It also detected de novo variants that occur in the offspring but are not present in either of the parents.

Patients with undiagnosed genetic disease and heterogenous phenotypes

Technology- NGS (Novaseq)

Specimen- whole blood, DBS card, CVS and DNA

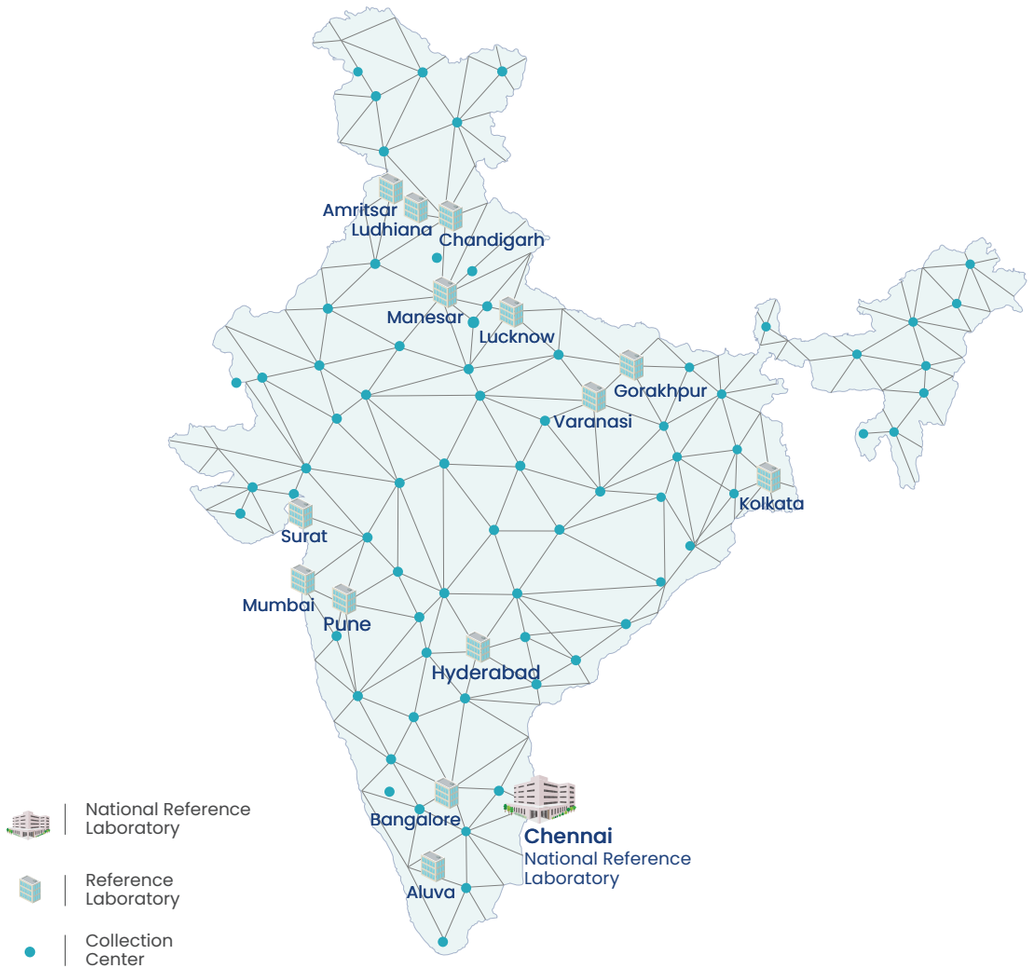
onco Life Exome panels

Test code	Test Name	Components	Technique	TAT	MRP
GEN0245_ONCO	Clinical Exome Sequencing(CES)_ONCO	CES	NGS	21 Days	18000
GEN0226_ONCO	Whole Exome Sequencing(WES)_ONCO	WGS	NGS	21 Days	25000
GEN0055_ONCO	Microdeletion and Duplication Analysis by MLPA_ONCO	Cri-du-Chat syndrome, Sotos syndrome, Saethre-Chotzen syndrome, Williams-Beuren syndrome, Williams-Beuren duplication syndrome, Langer-Giedion syndrome, WAGR syndrome, Prader-Willi/ Angelman syndrome, Rubinstein-Taybi syndrome, Miller-Dieker syndrome, Lissencephaly-1, Smith-Magenis syndrome, Potocki-Lupski syndrome, Alagille syndrome, DiGeorge syndrome, 22q11.2 microduplication syndrome, Phelan-McDermid syndrome	MLPA	10 Days	4,500
GEN0173_ONCO	Sanger Sequencing Confirmation_ONCO	Sanger confirmation of reported mutation	Sanger sequencing	15 Days	8,000
GEN0034_ONCO	Chromosomal Microarray(CMA 750 K) - Affymetrix Cytoscan 750K_ONCO	CMA	Affymetrix- CNV based analysis	8 Days	21000
GEN0632_ONCO	Clinical Exome Sequencing(CES) + Mitochondrial Genome Sequencing_ONCO	CES+ Mitochondrial Genome Sequencing	NGS	21 Days	20000
GEN0228_ONCO	Whole Exome Sequencing(WES) + Mitochondrial Genome Sequencing_ONCO	Whole Exome Sequencing(WES) + Mitochondrial genome	NGS	21 Days	27000

FAST GROWING NATIONWIDE

Care Delivery Network

LifeCell Diagnostics is now mfine /diagnostics



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Quality Reports

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- Hamilton NGS Star for High Precision Automation
- Fastest TAT for Maternal Markers
- Reproductive Genetics
- Karyotyping & CMA
- Oncology & Transplant
- Prenatal Screening of Aneuploidies
- Infectious
- Data and Academic Services
- Routine Diagnostics
- Fetal Autopsies

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