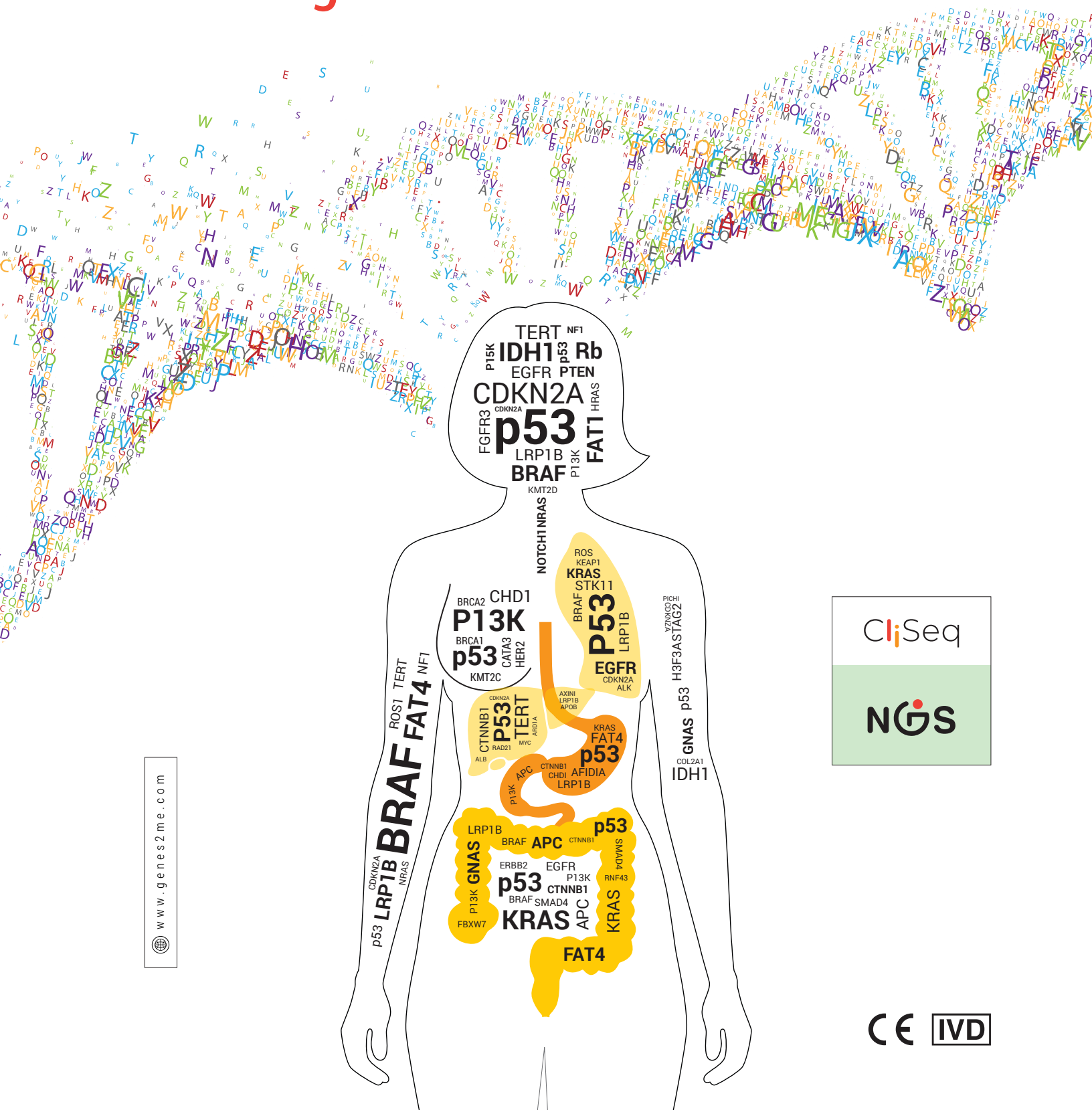
PanCan

G2M

Cancer Genomic Profiling



CliSeq

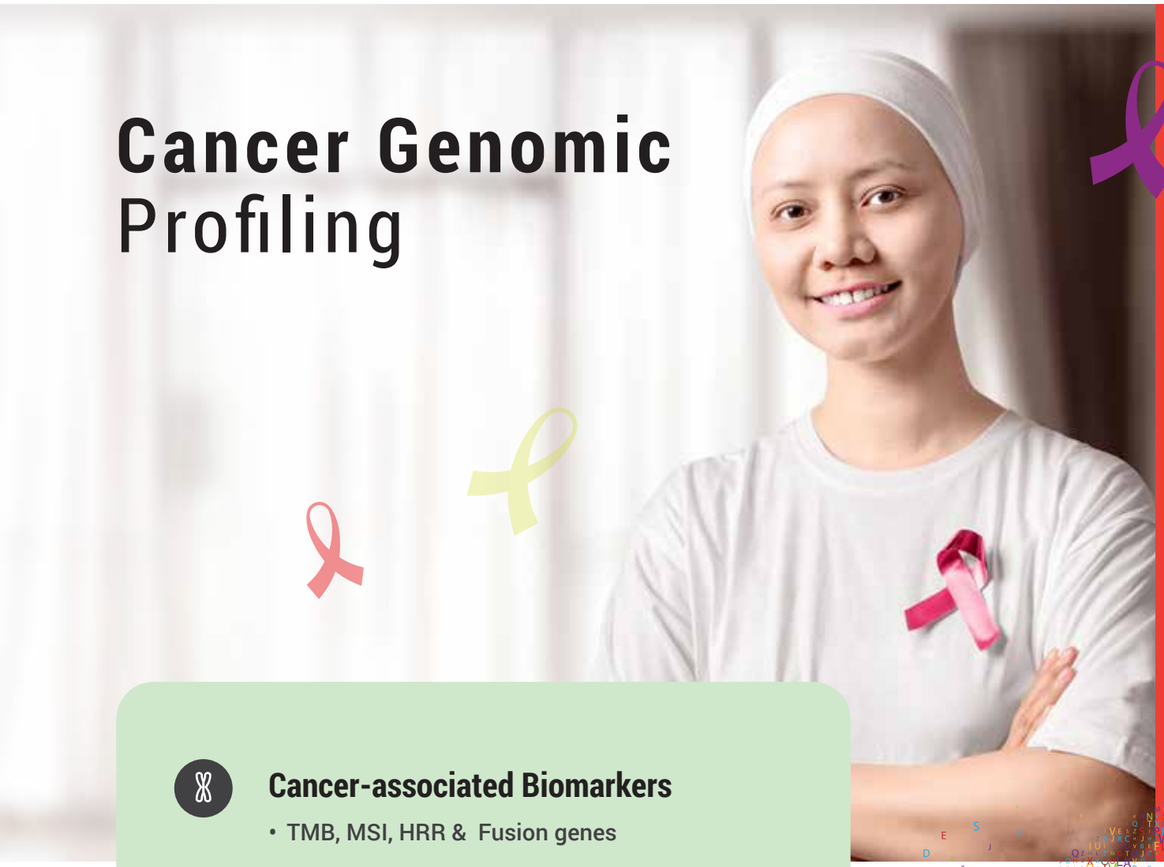
NGS

www.genes2me.com

CE IVD

PanCan | NGS

Cancer Genomic Profiling



Cancer-associated Biomarkers

- TMB, MSI, HRR & Fusion genes

- Uniform Depth Coverage
- Best On-Target Ratio
- Low Bias Base Call
- Less duplication rates
- Short Hybridization time of approx 4 hours
- Rigorously engineered to target hard to capture regions

PAN Cancer Panels

PAN Cancer Panels screens all variant types & immuno-oncology markers (MSI and TMB), which are crucial biomarkers for cancer immunotherapy

Pan Cancer Panel is an NGS panel aimed to screen a range of cancer causing genes to identify somatic mutations in DNA & RNA fusion from human research samples like FFPE and fresh tissue targeting 681 genes covering all the coding sequences enriched by Hybridization capture-based target enrichment.

Genes are selected based on AMP/ASCO/CAP guidelines to uncover the coding region compiling to the size of ~1.7 Mb. The genomic DNA & RNA from the FFPE or fresh tissue is considered for library preparation and enrichment that further can be sequenced on NGS sequencer. This panel detects all variant types and immune-oncology markers (TMB, MSI, HRR) which are crucial for cancer immunotherapy.

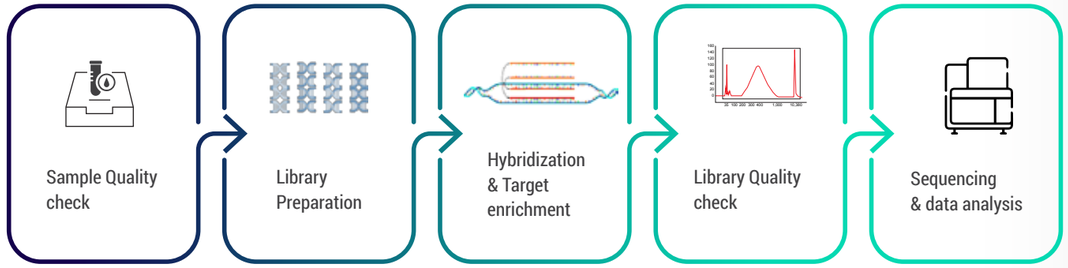
This panel covers major mutation types like SNVs, Indels, CNV and covers the DNA and RNA fusions as well. Results of the assay are used as an aid in identifying related cancer variants in combination with a patient's clinical indications. The panel is also designed to detect Epstein-Barr virus (EBV) & Human Papilloma Viruses (HPV), allowing for the comprehensive analysis of cancer-associated genes

No. of Genes	681
Gene Count / Family	524
Covered Region	Whole CDS, Hotspots, Fusion Genes
Target Size	~1.7 Mb
Mutation Type	SNV/ InDels / CNV
Biomarkers	TMB, MSI, HRR
Sample Type	FFPE & Fresh Frozen Tissue

PROCESS WORKFLOW

Platform Agnostic

Compatible on multiple platforms (Illumina, Thermo Fisher, Element Biosciences, MGI)



Bioinformatics Solution

Data Analysis and Interpretation using Genes 2Me Cliseq Interpreter plaftform

After raw data generation, we follow the GATK best practices framework for identification of variants in the sample, starting with raw data quality check using the FastQC followed by BWA read aligner for mapping/aligning to human reference genome GRCh38. After the alignment, GATK Mutect2 algorithm is used for variant calling. Annotation of the variants is performed using open-source available software SnpEff.

Further relevant mutations are annotated using published variants in literature and set of diseases databases – ClinVar, OMIM, COSMIC and HPO. The 1000Genome, gnomAD, dbSNP databases are used for annotation of variants for their minor allele frequency. The dbNSFP database is used for annotation and functional prediction of all potential non-synonymous variants.

CliSeq Interpreter

Sequencing Data Analysis Software

- Cloud based (CPU) analysis Software
- Global Software Access
- Strong customer Data Security

PANEL PERFORMANCE

Features	Illumina	MGI	Thermo Fisher
Coverage uniformity	>98%	>98%	>84%
Precision	>95%	>96%	>89%
Reproducibility	97%	97%	95%
Sensitivity	1% *VAF @ 95%	1% VAF @ 95%	1% VAF @ 95%
On Target Ratio	86-95%	85-95%	74-85%

*VAF - Variant Allele Frequency

SAMPLE REPORT

Accession ID:	390_3
Sample Type:	Blood + FFPE
Patient's Name:	PAN-CAN-TEST
Gender/Age:	Male/43 Years

PanCan
PAN CANCER PANEL

Data Uploaded On:	10/08/2023	Report Generated On:	08/09/2023

CANCER TYPE:

breast cancer

RESULTS:

List of clinically significant mutations (CSM)

Variants of potential clinical significance:

GENE	VAF	FDA Approved Drugs	
		Responsive	Resistance
BRAF (p.Pro403LeufsTer8)	8.73%		

Technical Information:

Variant	Depth	Genomic location	VAF
BRAF(NM_004333.6):c.1208del	Total: 1019 Alt: 89	chr7:140783126	8.73
Gene summary: Serine/threonine-protein kinase B-raf (BRAF), is a member of the Raf family of serine/threonine protein kinases, which signals through the MAP kinase pathway to regulate cell proliferation and cell growth. BRAF mutations and fusions have been identified in a variety of cancers, including melanoma, non-small cell lung cancer, colorectal cancer, papillary thyroid cancer, and ovarian cancer, and a number of mutations have also been demonstratied to confer drug resistance. BRAF fusion with TIF1A is observed in acute promyelocytic leukaemia, with SND1 in lung adenocarcinoma and with TRIM4 in lung invasive mucinous adenocarcinoma			

Therapeutic Summary:

Drug	Biomarker	Resistance/Responsive	Level	Clinical Indication

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Recommendations
• Genetic counseling is advised for interpretation on the consequences of the variant(s). • If results obtained do not match the clinical findings, additional testing should be considered as per referring clinician's recommendation. • Genetic test results are reported based on the recommendations of Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists • Sanger validation is recommended for the above listed variants. • The significance/classification of the variant(s) may change based on genetic testing in the parents and other family members. • Data revaluation performed upon the up gradation of databases used and results may vary in accordance.

AMP-ASCO-CAP CLASSIFICATION CRITERIA	Methodology
Four-tiered system to categorize somatic sequence variations based on their clinical significance 1. Tier I - Variants of strong clinical significance. 2. Tier II - Variants of potential clinical significance. 3. Tier III - Variants of unknown clinical significance 4. Tier IV - Benign or likely benign variants	The Genes2Me Pan Cancer NGS panel was used for sequencing that screens for 524 clinically relevant genes (coding regions of the genome) for diseases associated with genetic mutations. It covers all major mutations like SNV, InDels, CNV & other biomarkers such as MSI & TMB adding up to a target size of 2.5Mb with hybridization-based target capture technique. After raw data generation, we follow the GATK best practices framework for identification of variants in the sample, starting with raw data quality check using the FastQC followed by BWA read aligner for mapping/aligning to human reference genome GRCh38. After the alignment, GATK Mutect2 algorithm is used for variant calling. Annotation of the variants is performed using open-source available software SnpEff. Further, clinically relevant mutations are annotated using published variants in literature and set of diseases databases – ClinVar, OMIM, COSMIC and HPO. The 1000Genome, gnomAD, dbSNP databases are used for annotation of variants for their minor allele frequency. The dbNSFP database is used for annotation and functional prediction of all potential non-synonymous variants.

Limitations	Disclaimer
• Genetic testing is an important part of the diagnostic process however it may not always give a definitive answer. In some cases, a genetic variant may be missed due to the limitations in existing medical knowledge and testing technology. Accurate interpretation of test results is dependent on the availability of biological & medical information (clinical history) of the family, failing to this may leads to incorrect result interpretation and diagnosis. • Test results are interpreted in the context of clinical findings, available scientific evidences, family history and other laboratory data. The	• The interpretation of variants in this report is performed to the best knowledge of the scientific & medical information available at the time of reporting. The classification of variants is based on AMP-ASCO-CAP guidelines. • As of the inherent technological limitations of the sequencing assay, some of the coding regions can't be properly sequenced, so, variations in these regions may not be identified & interpreted. It is possible that variants not identified by the assay may be associated with the provided phenotypes of the patient.

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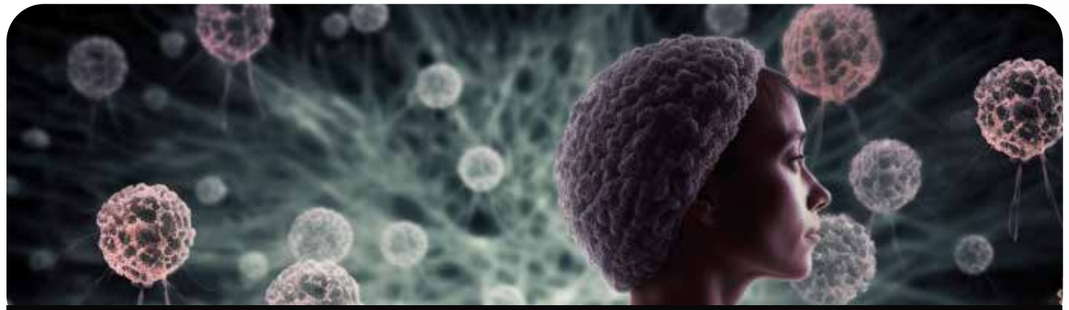

COVERS WIDE RANGE OF CANCERS*

Colorectal Cancer
- →RAS
- →EGFR
- →TGF
- →ATM
- →BRAF
- →CHEK2
- →NRAS
- →PIK3CA
- →PTEN
- →TP53

Breast Cancer
- →BRCA 1
- →BRCA 2
- →PTEN
- →TP53
- →CHEK2
- →BRIP1
- →ATM
- →PALB2
- →PIK3CA
- →PMS2
- →ESR1
- →FGFR1

Ovarian Cancer
- →CHEK2
- →TP53
- →BARD1
- →KRAS
- →RAD51
- →BRIP1
- →PALB2
- →BRAF
- →ERBB2
- →PTEN
- →PIK3CA
- →BRCA1

Bladder Cancer
- →TP53
- →RB1
- →HRAS
- →PIK3CA
- →FGFR3
- →ATM
- →MTOR

Lung Cancer
- →EGFR
- →KRAS
- →ALK
- →RET
- →PIK3CA
- →EGFR
- →KRAS
- →MET
- →PTEN
- →RET
- →BRAF
- →ERBB2
- →ALK

Oesophagus Cancer
- →ERBB2
- →EGFR
- →RB1

Prostate Cancer
- →BRCA 1
- →HOXB13
- →AR
- →ATM
- →MYC
- →PTEN
- →RAF1
- →BRCA2

Pancreatic Cancer
- →BRCA 1
- →BRCA 2
- →EGFR
- →HRAS
- →KRAS
- →PALB2
- →PIK3CA
- →TP53

Thyroid Cancer
- →BRAF
- →RAS
- →RET
- →TP53
- →PTEN

Cervical Cancer
- →DICER1
- →MED1
- →HLA-A
- →PI3K
- →MAPK

Liver Cancer
- →TP53
- →CDKN2A

Gastric Cancer
- →APC
- →MLH1
- →MSH2
- →MSH6
- →EPCAM

Limited Cancer Gene list presented here

The Genes2Me Pan Cancer Panel screens broad range of cancer causing genes to identify somatic mutations in the tumor tissue.

It provides comprehensive detail of the cancer and recommendations regarding the FDA approved drugs, helps to decide the best course of treatment.

Scan for PanCan Gene List

Gene & Drug Recommendations

TYPE OF CANCER*	GENE	DRUG
Gilioma, Acute Myeloid Leukemia	IDH1	Olutasidenib
Breast Cancer, Ovarian Cancer	BRCA1	Olaparib
NSCLC, Colorectal Cancer	EGFR	Osimertinib
Colorectal Cancer, NSCLC	KRAS	Cetuximab
NSCLC, Melanoma, Metastatic Colorectal Cancer	BRAF	Encorafenib
Follicular Lymphoma Tumor	EZH2	Tazemetostat
Medullary Thyroid Cancer, Thyroid Cancer	RET	Selpercatinib
Prostate Cancer	BRCA2	Niraparib
Breast Cancer, Gastroesophageal Cancer	ERBB2	Trastuzumab
Non-Small Cell Lung Cancer	ALK	Alectinib
Esophageal, colorectal, Lung cancer	TP53	Venetoclax
Breast Cancer, Ovary, stomach cancer	PIK3CA	Alpelisib
Gastrointestinal Stromal Tumors, glioblastoma, melanoma	PDGFRA	Avapritinib
Urothelial Cancer, multiple myeloma, bladder cancer	FGFR3	Erdafitinib
NSCLC, Metastatic cancer	MET	Capmatinib
Myeloma , lung adenocarcinoma, colon adenocarcinoma, melanoma, breast carcinoma	PDGFRB	Imatinib Mesylate
Acute Myelogenous Leukemia, Bone Marrow cancer	FLT3	Quizartinib
Aggressive Systemic Mastocytosis, lung adenocarcinoma,	KIT	Imatinib
colon adenocarcinoma	ESR1	Elacestrant
Breast Cancer, endometrial and prostate cancer Solid Tumors, lung cancer, colorectal cancer	NTRK1	Entrectinib

Limited cancer type details mentioned

ORDERING INFORMATION

Commercial Name	Cat No.
PAN Cancer Panel	G2MPC06001-ill; G2MPC06001-TF; G2MPC06001-MG

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Genes2Me Pvt. Ltd.

1105, 11th Floor, Tower B, SAS Tower, Medicity,
Sector - 38, Gurgaon - 122 001, Haryana, India

Tel : + 91 18001 214030 / + 91 88000 23600
E-mail : contact@genes2me.com

www.genes2me.com