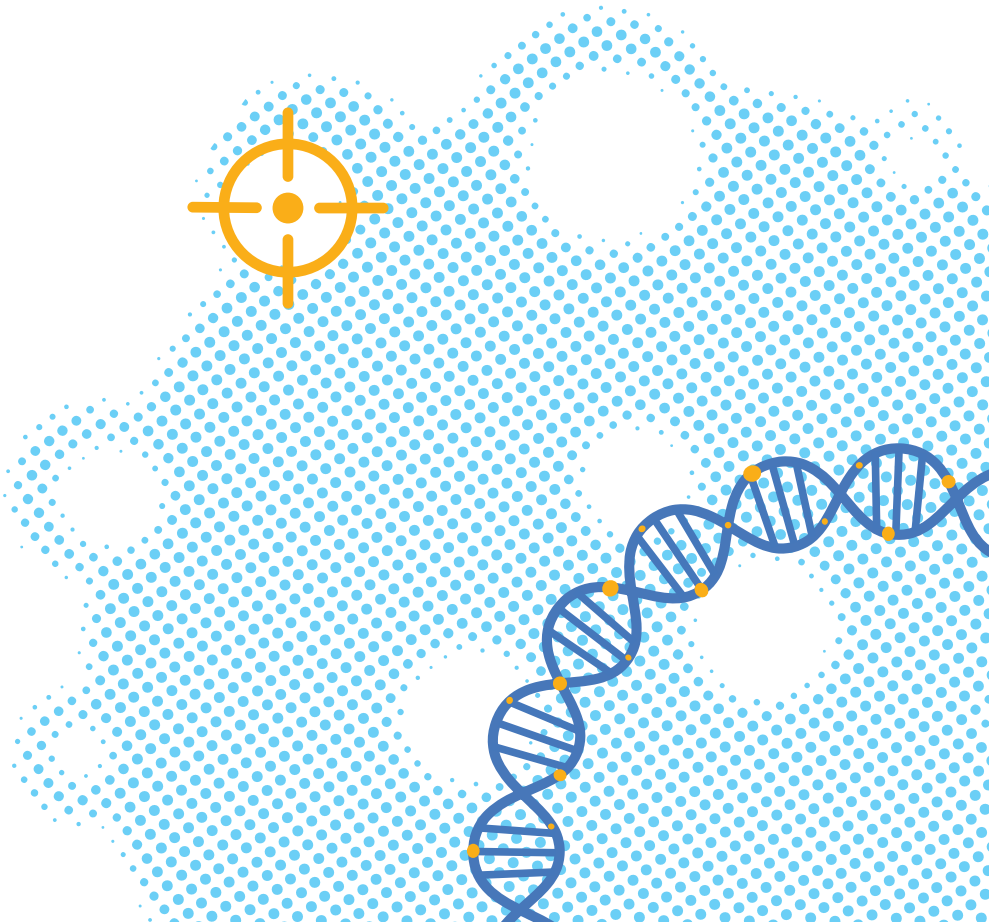
Prima
Cancer Genetics

MEDGENOME

Somatic Cancer Mutation Panel



Cancer in India

There are around 2.5 million people estimated to be living with cancer

For every 2 women newly diagnosed with breast cancer, one woman dies of it in India

As many as 2,500 people die every day due to tobacco-related diseases

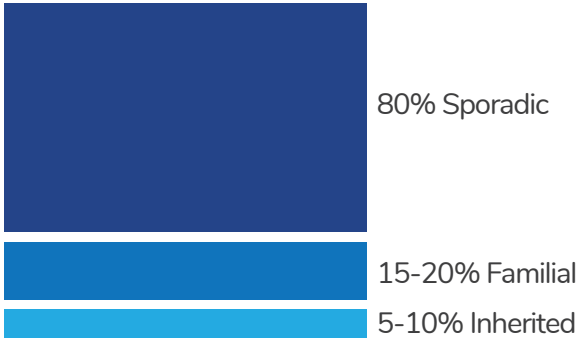
Cancers of oral cavity and lungs in males and of cervix and breast in females account for over 50% of all cancer deaths

Top 5 Cancers in Men and Women in India

MEN	WOMEN
Head & Neck	Breast
Lung	Cervix
Gastric	Colorectum
Colorectum	Ovary
Pharynx	Oral Cavity

Top 5 Cancers in Men and Women in India

In its projection, the Indian Council of Medical Research (ICMR) said in 2016. “India is likely to have over 17.3 lakh new cases of cancer and over 8.8 lakh deaths due to cancer by 2020 with cancers of breast, lung and cervix topping the list”.



What are Acquired or Somatic mutations?

- Most cancers are caused by acquired mutations
- These mutations cannot be inherited
- Do not occur in reproductive cells (egg or sperm cells)
- Are much more common than inherited mutation
- An individual with a germline mutation may also develop a somatic mutation

Advantages of tumor DNA analysis

Discovery and knowledge of somatic mutations in a cancer:

- Enables molecular targeted treatment
- Provides clues to the underlying biology of cancer
- Elucidates the primary genetic changes driving tumorigenesis
- Provides new molecular drug targets

When to suggest for the somatic mutation panel?

- Patients with solid tumors
- Patients with aggressive solid tumors
- Patients with metastatic solid tumors
- Patients with solid tumors who have failed first-line therapy or who are non-responsive to first-line therapy

Next Generation Sequencing and its benefits

- NGS panels are tests that analyse multiple genes simultaneously
- It is done at a much lower cost than traditional sequencing methods
- Has a higher likelihood of identifying a causative mutation
- Reduces the need for multiple follow-ups and additional testing

Next Generation Sequencing (NGS) at MedGenome

Using genomic DNA extracted from FFPE tumor tissue, the coding regions of all the genes are captured and sequenced simultaneously by NGS technology on an Illumina platform. The sequence data that is generated is aligned and analyzed for sequence variants.

SOMATIC Cancer Mutation (hotspot tumor) panel

Detects hotspot mutation in 56 cancer-related genes

ABLI, AKTI, ALK, APC, ATM, BRAF /inclusive of v600E), CDHI, CDKN2A, CSFIR, CTNNBI, DDR2, DNMT3A, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFRI, FGFR2, FGFR3, FLT3, FOXL2, GNAII, GNAQ, GNAS, HNFIA, HRAS, IDHI, IDH2, JAK2, JAK3, KOR, KIT, KRAS, MAP2KI/MEK1, MET, MLHI, MPL, MSH6, NOTCHI, NPMI, NRAS, PDGFRA, PIK3CA, PTEN, PTPNII, RBI, RET, SMAD4, SMARCBI, SMO, SRC, STKII, TP53, TSCI, VHL

EX: Cancers of the Lung, Brain, Kidney, Colon, Stomach, Liver, Pancreas etc.

Sample Requirement

5-10 µm thickness from the FFPE tumor tissue accompanied with pathology report and oncology report in case of a relapse testing.

5-10 µm thickness from the FFPE tumor tissue

pathology and oncology report

How long does it take to get the results?

It will take approximately 4-5 weeks to complete the test and give the results.
Enables molecular targeted treatment

Free Genetic Counselling

Prima offers all your patients FREE pre & post test genetic counselling with our expert and Certified Genetic Counsellors.

Best available support for your patients and families via

- Latest technologies
- Helpful customer service
- Clear result interpretation
- Counselling sessions with our Genetic Counsellors

Sample Report

MEDGENOME

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#258/A, Bommasandra, Hosur Road, Bangalore – 560 099, India.
Tel : +91 (0)80 67154931/32/33/84 Web: www.medgenome.com

DNA TEST REPORT – MEDGENOME LABORATORIES

Patient Name	ABCD	Gender / Age	M / 73Y
Hospital Name	CDFG Hospital, Hyderabad	Unique Identification Number (UID)	XYZ
Physician Name	Dr. GHIJ	Sample Type	FFPE blocks [S-5050/16 (IB & IK)]
Test requested	Somatic Cancer Mutation Panel	Collection date	10-02-2018,10:00:00
		Received date	13-02-2018,12:53:32
MG Samples ID/ Order ID	3333/ 55555	Report Date	21-02-2018, 11:00:00

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Malignant Melanoma of anal canal. The tumor was identifiable in the blocks [S-5050/16 (IB & IK)] and it was adequate for further analysis.

RESULTS

No clinically relevant alterations that can be targeted with approved drugs have been identified

TABLE 1: GENOMIC ALTERATIONS THAT CAN BE TARGETED WITH APPROVED DRUGS IN THE SUBJECT'S TUMOR TYPE

Gene	CDS variant#	Amino acid variant / Exon No.	Overall Depth / Mutant Allele (%)	FDA Approved drugs	Drug response	Hot spot Mutation	Function of the gene in cancer
None							

TABLE 2: NON-DRUGGABLE/DRUGGABLE CLINICALLY SIGNIFICANT GENOMIC ALTERATIONS INDICATED IN OTHER TUMORS

Sl. No.	Gene	CDS variant#	Amino acid variant / Exon No.	Overall Depth/ Mutant Allele (%)	Impact on Protein Function	Function of the gene in cancer	Pathway in which the gene functions
1	***KIT***	c.1706_1722delTTTACATAGACCCAACAinsGGTCT (ENST00000288135])	p.Val569_Thr574delinsGly Ser / Exon 11	**11515X / 11%**	NA	Oncogene	MAPK and PI3K

TABLE 3: VARIANTS OF UNKNOWN SIGNIFICANCE

Sl. No.	Gene	CDS variant#	Amino variant / Exon No.	Overall Depth/ Mutant Allele (%)	Function of the gene in cancer
None					

CLINICAL CORRELATION AND VARIANT INTERPRETATION

***KIT* (p.Val569_Thr574delinsGlySer) (Table 2):**

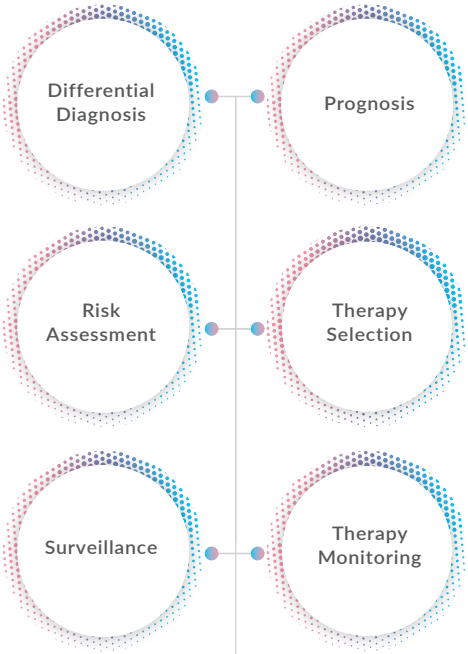
The *KIT* gene encodes a transmembrane receptor tyrosine kinase (RTK) protein and somatic activating mutations in *KIT* have been associated with various neoplasms, mainly Gastro-Intestinal Stromal Tumors (GIST), leukemias, malignant melanoma and thymic carcinomas. *KIT* is one of the major targets of Imatinib and mutations of *KIT* gene predict the treatment efficacy in GIST. Adjuvant imatinib significantly improves relapse free survival for patients with exon 11 mutated GIST, HR 4.85 (95% CI 1.49–15.76) [1].

SAMPLE REPORT

MEDGENOME

Prima by MedGenome offers a wide range of Oncology and Haematology genetic tests, these include:

Molecular Testing for Hematological Malignancies, Comprehensive Leukemia Panel



IGHV Gene Mutation Testing for CLL, Comprehensive Leukemia Panel, BCR-ABL1 gene fusion analysis

Hereditary Cancer Panel, BRCA1 and BRCA2 gene test, Thalassemia Mutation Test

Comprehensive Tumor Gene Panel, Somatic Mutation Panel, Comprehensive Leukemia Panel, Molecular Testing for Lung Cancer

OncoTrack, OncoSelect, OncoFocus (Liquid Biopsy Test)

NGS based IRMA, BCR- ABL1 gene fusion analysis

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COLLEGE of AMERICAN PATHOLOGISTS

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