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# SOLID TUMOR NEXT-GENERATION SEQUENCING (NGS) PANELS:

The NGS panels described below are all run on the lonTorrent platform. The genes included in each panel are known to be frequently mutated in corresponding neoplasms and malignancies, and their presence may affect diagnosis, prognosis and/or treatment modalities. In addition to mutation calls, eligible clinical trial information, when available, is provided with our multigene panels.

#### **50 GENE CANCER PANEL**

Currently our largest panel, this assay has been validated to identify hotspot mutations in the listed genes using **formalin fixed**, **paraffin embedded tissues**.

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

#### **DISEASE SPECIFIC SUB-PANELS**

These smaller, less costly panels, test for the most common hotspot mutations found in specific tumor types. Each of the sub-panels is run using the same technology platform as the 50-Gene Cancer Panel.

#### **GASTROINTESTINAL STROMA TUMOR (GIST)**

		1
BRAF	KIT	PDGFRA

#### **COLON CANCER MUTATION PANEL**

BRAF	HRAS	KRAS	NRAS	PIK3CA

#### LUNG CANCER MUTATION PANEL - WITHOUT FISH FOR ALK/ROS1

AKT1	EGFR	ERBB4	NRAS	PTEN
BRAF	ERBB2	KRAS	PIK3CA	

#### **MELANOMA MUTATION PANEL**

BRAF GNA11 GNA	S HRAS	KIT NRAS
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# MYELOID NEOPLASM NEXT-GENERATION SEQUENCING PANELS:

# **MYELOID MUTATION PANEL**

This assay identifies somatic mutations in 40 genes that are known to be mutated in myeloid malignancies including Acute Myelogenous Leukemias (AML), Myelodysplastic Syndromes (MDS) and Myeloproliferative Neoplasms (MPN). Information regarding the diagnostic, prognostic and/or therapeutic significance of variant(s) will be reported, when applicable, and findings by NGS will be integrated with findings from other molecular assays, FISH and/or karyotype, when applicable and available from additional testing. This testing uses **non-formalin fixed tissues**. Shaded boxes designate full gene coverage.

ABL1	CALR	DNMT3A	GATA2	IKZF1	MPL	NRAS	RB1	SH2B3	TP53
ASXL1	CBL	ETV6	HRAS	JAK2	MYD88	PHF6	RUNX1	SRSF2	U2AF1
BCOR	CEBPA	EZH2	IDH1	KIT	NF1	PRPF8	SETBP1	STAG2	WT1
BRAF	CSF3R	FLT3	IDH2	KRAS	NPM1	PTPN11	SF3B1	TET2	ZRSR2

# **MYELOPROLIFERATIVE NEOPLASM (MPN) SUB-PANEL**

This is a smaller panel derived from the myeloid mutation panel with a focus on the genes most commonly mutated in myeloproliferative neoplasms (e.g. polycythemia vera, essential thrombocythemia, myelofibrosis).

CALR	CSF3R	JAK2	KIT	MPL
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# **NEXT-GENERATION SEQUENCING SINGLE GENE ASSAYS**

Single gene mutation analysis is also available on select genes listed below.

If considering more than one separate gene test, we highly recommend ordering one of the panels from above. Trial information is NOT included with single gene assays.

Solid Tumor

•BRAF

•KRAS •IDH1

•GNAS

- PIK3CA
  - •TP53
- •EGFR •CTNNB1

- Myeloid Neoplasm
- •CEBPA •MYD88
- •FLT3 (TKD) •KIT
- •IDH1/2 •TP53
- •JAK2 (Exon 14 Only)
- •JAK2 (Exons 12-15; Polycythemia Vera)
- JAK2 (V617F\_Reflex to Exon 12)



# **MISCELLANEOUS TESTING OFFERED**

#### Molecular for Solid Tumor

- Microsatellite instability (MSI) by Capillary Electrophoresis: Separate Tumor and Normal tissue required
- Synovial Sarcoma RT-PCR
- BRAF p.V600E by Pyrosequencing
- MGMT Methylation Status for Gliomas
- Ewings Sarcoma
- Rhabolomyosarcoma

#### Molecular for Hematologic Malignancy

- T-Cell Gene Rearrangement
- B-Cell Gene Rearrangement • FLT3-ITD by Capillary
- Electrophoresis
- NPM1 Insertion by Capillary Electrophoresis
- PML-RARA Rearrangement by RT-PCR

• BCR/ABL QuaLitative: (For

• BCR/ABL QuaNtitative: (For

increased sensitivity)

characterizing rearrangements,

monitoring established disease

- Ki-67
- MSI testing)PD-L1

# Immunoperoxidase Staining and Interpretation

Listed is a small subset of stains commonly used in assessing tumor prognosis and for predicting response to therapeutics.

- ALK
- AR (androgen receptor)
- ER
- PR
- Her2/neu
- MMR (Mismatch repair enzyme status [MLH1, PMS2, MSH2, MSH6], a surrogate for

Polycythemia Vera)

• KIT (Systemic Mastocytosis)

• NPM1

TP53

# FLUORESCENCE IN-SITU HYBRIDIZATION (FISH) TESTING

status)

Send out FISH testing is available for a variety of soft tissue neoplasms and malignancies. Please refer to the table below.

	17 Centromere	19q13/19p13	1p36 deletion	1p36/1q25	3 Centromere	7 Centromere	ALK [2p23]	CDKN2A )p16) [9q21]	COL1A1/PDGFB[t(17;22)]	DDIT3 (CHOP) [12q13]	EGFR [7p12]	ETV6 (TEL) [12q13]	EWSR1 [22q12]	EWSR1/ATF1 [t(12;22)]	FUS [16p11.2]	FUS/CREB3L2 [t(7;16)]	HMGA2 [12q14.3]	MYC [8q24]/8Centromere	MYCN [2p24.1]	NUTM1 [15q14]	NUTM1/BRD4 [t(15;19)]	PLAG1 [8q12.1]	PTEN [10q23]	SMARCB1 9INI1) [22q11.23]	SS18 (SYT) [18q21/t]	TFE3 [Xp11]	USP6 [17p13]
Bladder Cancer																											
Clear Cell Sarcoma of Soft Tissue (CCS)																											
Dermatofibrosarcoma Protuberans (DFSP)																											
Ewing Sarcoma (ES)/ Primitive Neuroectodermal Tumor (PNET)																											
Extraskeletal Myxoid Chondrosarcoma																											
Infantile Fibrosarcoma (IFS)																											
Fibroblastic Tumor (IMT)																											
Lipoblastoma																											
Lipoma																											
Liposarcoma (WDLS/ALD; DDLS)																											
Mammary Analogue Secretory Carcinoma (MASC)																											
Medulloblastoma (MBD)																											
Myxoid Liposarcoma (MLS)																											
Neuroblastoma (NB)																											
Neurological Cancer																											
Nodular Faciitis																											
Phabdoid Tumor (AT/RT; MRT)																											
Synovial Sarcoma (SS)																											

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# Every patient's cancer is unique... So is our approach to diagnosis

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