

# CAPN

Solid Tumor Targeted Cancer Gene Panel by Next Generation Sequencing

Patient ID SA00000770	Patient Name SAMPLEREPDMGLM, VLD20150713	Birth Date 1981-01-01	Gender M	Age 34	
Order Number SA00000770	Client Order Number SA00000770	Ordering Physician Client, Client	Report Notes	,	
Account Information C7028846 DLMP Rochester		Collected 12 Jul 2015 13:57			

**Result Summary** 

MCR

# **Alteration Identified**

Result									MCR
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	
CNAO	CDC	CTI/11	TDEO	VIII					

Provided diagnosis: colorectal adenocarcinoma

No additional reportable somatic alterations were identified within the tested genes.

The following alteration was identified:

Gene: KRAS

DNA change: c.35G>A

Amino Acid change: p.G12D (Gly12Asp)

Classification: MUTATION

# Interpretation

1 MCR

# ASSOCIATIONS BETWEEN KRAS MUTATIONS AND COLORECTAL CANCER

Approximately 35% of patients with colorectal adenocarcinoma have a somatic mutation in the KRAS gene (1). KRAS mutations, primarily those occurring at codons 12, 13, and 61, result in constitutive activation of the RAS/MAPK signaling pathway.

Current data suggests that the efficacy of EGFR-targeted therapies in colorectal cancer is limited to patients with tumors lacking KRAS mutations. Thus, the detection of a KRAS activating mutation within this tumor suggests that EGFR-targeted therapies may have limited therapeutic value for this patient (2).

# **REFERENCES**

- 1. http://cancer.sanger.ac.uk/cancergenome/projects/cosmic/
- 2. Ann Oncol. 2013 Aug;24(8):2062-7 (PMID 23666916)

# ADDITIONAL INFORMATION

Microscopic examination was performed by a pathologist to identify areas of tumor for enrichment by macrodissection. Next generation sequencing is performed to test for the presence of a mutation within targeted regions of the following genes: 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, and VHL.

Mutation nomenclature is based on build GRCh37 (hg19). For details about gene reference transcripts (GenBank accession

# **Performing Site Legend**

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905



Patient ID	Patient Name		Birth Date	Gender	Age
SA00000770	SAMPLEREPDMGLM, VLD20150713A0029		1981-01-01	M	34
Order Number SA00000770	Client Order Number SA00000770	Ordering Physician Client, Client	Report Notes		
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numbers) and additional information about this test, see www.mayomedicallaboratories.com (Test ID CAPN).

#### **CLINICAL CORRELATIONS**

Test results should be interpreted in context of clinical findings, tumor sampling, histopathology, and other laboratory data. If results obtained do not match other clinical or laboratory findings, please contact the laboratory for possible interpretation. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

The presence or absence of a mutation may not be predictive of response to therapy in all patients.

#### **TECHNICAL LIMITATIONS**

This test does not detect large insertions, deletions, or duplications or genomic copy number variants.

This assay has been shown to detect >99% of single base

substitutions and >93% of known COSMIC insertions and deletions up to 22bp in length within the reportable range of this assav.

A negative (wild type) result does not rule out the presence of a mutation that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay is 5–10% with a minimum coverage of 100X.

Rare polymorphisms may be present that could lead to false negative or false positive results.

This test cannot differentiate between somatic and germline alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

Metastatic and corresponding primary lesions may have discordant results.

Additional Information

#### **CLINICAL TRIALS**

Possible clinical trials of benefit for this patient can be found at the following sites:

1) ClinicalTrials.gov:

http://clinicaltrials.gov/ct2/search/advanced

2) Mayo Clinic:

http://www.mayo.edu/research/clinical-trials/

3) National Cancer Institute:

http://www.cancer.gov/clinicaltrials/search

# REFERENCE TRANSCRIPT(S)

Mutation nomenclature is based on the following GenBank accession number(s) (build GRCh37 (h19)): KRAS NM\_004985.

**Specimen** 

MCR

Tissue, Tumor

Tissue ID

S15-5999

Released By

EMILY LAUER

**Received:** 13 Jul 2015 20:19 **Reported:** 24 Jul 2015 10:50

#### **Laboratory Notes**

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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