Patient Name

Age/Gender

OP/IP No/UHID

MaxID/Lab ID

Ref By

Centre

OP/IP No/UHID

Collection Date/Time

Reporting Date/Time



# **TEST REQUESTED**

Max Oncomine myelodysplastic syndrome (MDS) panel

## **CLINICAL INFORMATION**

Hypercellular bone marrow showing disproportionate decrease in erythroid precursors viz-a-vis the degree of anaemia with absent iron stores and mild dyshemopoietic changes.

#### **TARGETED GENES**

HOTSPOT GENES COVERED (Next Generation Sequencing)													
CBL	DNMT3	A FLT.	FLT3 (		GATA2 HF		IRAS			IDH2	JAK2	KIT	KRAS
MPL	MYD88	NPI	NPM1		RAS PTF		SF3B1		SRSF2		U2AF1	WT1	
FULL GENES COVERED (Next Generation Sequencing)													
ASXL1	BCOR	CALR	ALR CE		BPA ETV6		EZHZ	2 IKZF1		?F1	NF1	PHF6	PRPF8
RB1	RUNX1	SH2B3	2B3 ST		AG2 TET2		TP53		ZRSR2				
FUSION DRIVER GENES COVERED (Next Generation Sequencing)													
ETV6	JAK2	KMT2 (MLL)		MECON	1	МЕТ	-	MLLT	10	MLLT3	RARA	RUNX1	

# **PRIMARY FINDINGS**

No Pathogenic variant found No Fusion found

# **INTERPRETATION SUMMARY**

- This test did not identify any clinically significant variant in the genes mentioned in the panel.
- This test did not identify any fusions in the genes mentioned in the panel.

SAMPLE STATISTICS					
Coverage	99.62%				
Depth	2,518				

Patient Name

Age/Gender

OP/IP No/UHID

MaxID/Lab ID

Ref By

Centre

OP/IP No/UHID

Collection Date/Time

Reporting Date/Time



#### **TEST METHODOLOGY**

### **Background**

Multi gene analysis through next generation sequencing allows the identification of variants to understand their prognostic and therapeutic implications in different cancer types, if any. Targeted application of next-generation sequencing (NGS) technology allows detection of specific mutations that can provide treatment opportunities to the patients. This panel targets 40 key genes, 29 fusion driver genes and uses methodologies of Next generation sequencing using Oncomine myeloid assay. These genes have been selected on the basis of their known impact as actionable targets of existing and emerging anti-cancer therapies, and the prognostic features in specific tumor types. The sensitivity of the assays depends on the quality of the sample and tumor content.

#### Method

The Oncomine myeloid assay was used to carry out next generation sequencing. After sequencing, automated analysis was performed with Torrent Suite™ Software. Variant annotations were then done using Ion Reporter™ Software. Clinically relevant mutations were also checked using published literature and databases.

#### Limitations

The accuracy and completeness may vary due to variable information available in different databases. The classification of variants of unknown significance can change over time. Synonymous mutations were not considered while preparing this report. The mutations have not been confirmed using Sanger sequencing and/or alternate technologies.

### **DISCLAIMER**

A Negative result implying non-detection of mutation/deletion indicates a Benign/likely Benign polymorphism. A negative test result may also be due to the inherent technical limitations of the assay. Results obtained should be interpreted with consideration of the overall picture obtained from clinical, laboratory, and pathological findings. Rare polymorphisms may lead to false negative or positive results. False negative results may be due to sampling error/errors in sample handling as well as clonal density below the limit of detection. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Identification of a mutation in one or more of these genes does not guarantee activity of the drug in a given indication due to the presence of contraindicated mutation in the gene not covered by the panel.

The accuracy and completeness may vary due to variable information available in different databases. Classification of the variant may change overtime. An updated variant classification may be obtained on request. Insertions and deletions greater than 20bp in size may not be detected by this assay. The scope of this assay limits to SNVs, MNVs, short deletions/duplications and fusions. Due to poor quality of sample, indeterminate result due to low gene coverage or low variant depth cannot be ruled out.

The information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician. Patient care and treatment decisions should only be made by the physician after taking into account all relevant information available including but not limited to the patient's condition, family history, findings upon examination, results of other diagnostic tests, and the current standards of care. This report should only be used as an aid and the physician should employ sound clinical judgment in arriving at any decision for patient care or treatment.

(DR ATUL THATAI)
Director Molecular &
Cytogenomics

(DR NITIN DAYAL)
Prin. Cons. and Head
Hematopathology

Raw

(DR PRIYANKA AGARWAL) Sr. Manager Genomics & Molecular