Regd. Dt:	19/02/2018	Acc. ID: 27178865		Client Details:	Patient Care Centre (Chandigarh)
Coll Dt. Tm:	18/02/2018 00:00:00				SCO No. -140, Sector -240Chandigarh
Recd Dt. Tm:	19/02/2018 10:38:31			Refd. By:	C/O. KOS DIAGNOSTIC LAB
Age:	46 Yrs	Sex:	Male	Report Dt. Tm:	28/02/2018 16:26:14
Name:	Mr. RUPESH GAUR				

JAK2 Exon 12 Mutation Analysis (Qualitative) ^

PCR & Gene Sequence Analysis

Specimen type: EDTA P Bld

	RESULT _	
	IN RANGE	**OUT OF RANGE**
***JAK2* Exon 12 Mutation**	**Not detected**	**###**

Result:

No mutation was observed in exon 12 of *JAK2* gene in the specimen provided.

Interpretation:

Chronic myeloproliferative disorders (CMPD) are clonal hematopoietic stem cell-disorders characterized by proliferation of one or more myeloid cell lineages. CPMDs include polycythemia vera (PV), essential thrombocythemia (ET), idiopathic myelofibrosis (IMF), chronic myeloid leukemia (CML), hypereosinophilic syndrome (HES), chronic eosinophilic leukemia (CEL) and chronic neutrophilic leukemia (CNL). The primary characteristics of PV and ET are the increased production of RBC and platelet leading to the clinical manifestation of thrombosis or hemorrhage. A point mutation at residue 617 in exon 14 of *JAK2,* which arises from a G to T transversion and result in Valine to Phenylalanine change, has been reported in 70-85% cases of PV and 40-50% cases of ET and IMF. Many MPD cases negative for exon 14 mutations have been observed to carry mutations in exon 12 of *JAK2*. Somatic mutations in exon 12 of *JAK2* have been found in 5-15% cases of suspected PV.

Test Attributes and Limitations:

This assay is based upon PCR and Gene Sequencing of Exon 12 of *JAK2* gene(RefSeq NM_004972). The analytical sensitivity of the test allows detection of the mutation when the mutant clone comprises at least 18-20% of the total genomic DNA.
Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of high molecular weight DNA. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.

Note: This Test has been developed and its performance evaluated at Oncquest Laboratories Ltd.

B.Marrow/P Bld. EDTA

Calreticulin (*CALR)* Mutation Analysis (Qualitative) ^

PCR & Gene Sequence Analysis

Specimen type: EDTA P Bld

	RESULT _	
	IN RANGE	**OUT OF RANGE**
***CALR* Exon 9 Mutation(s)**	**Not detected**	*******

The sample is processed by Oncquest Laboratories Ltd.

CAP accredited **^ In Scope of NABL**

Regd. Dt:	19/02/2018	Acc. ID: 27178865		Client Details:	Patient Care Centre (Chandigarh)
Coll Dt. Tm:	18/02/2018 00:00:00				SCO No. -140, Sector -240Chandigarh
Recd Dt. Tm:	19/02/2018 10:38:31			Refd. By:	C/O. KOS DIAGNOSTIC LAB
Age:	46 Yrs	Sex:	Male	Report Dt. Tm:	01/03/2018 14:34:56
Name:	Mr. RUPESH GAUR				

Result:
No mutation was observed in exon 9 of *CALR* gene in the specimen provided.

Interpretation:
Chronic myeloproliferative disorders (CMPD) are clonal hematopoietic stem cell-disorders characterized by proliferation of one or more myeloid cell lineages. CMPDs include polycythemia vera (PV), essential thrombocythemia (ET), primary myelofibrosis (PMF), chronic myeloid leukemia (CML), hypereosinophilic syndrome (HES), chronic eosinophilic leukemia (CEL) and chronic neutrophilic leukemia (CNL).
CALR gene found at 19p13 encodes a calcium binding chaperone protein involved in glycoprotein folding and calcium homeostasis. A spectrum of insertion/deletion mutations have been observed in exon 9 of *CALR* gene in a subset of MPN patients who lack mutations in *JAK2* and *MPL* genes. *CALR* mutations are restricted to patients with ET and PMF and are not observed in those with PV. *CALR* mutated ET and PMF are associated with increased overall survival and decreased incidence of thrombosis.

Test Attributes and Limitations:
This assay is based upon PCR and Gene Sequencing of Exon 9 of *CALR* (RefSeq NM_004343). The analytical sensitivity of the test allows detection of the mutation when the mutant clone comprises at least 18-20% of the total genomic DNA.
Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of viable high molecular weight DNA. This is a diagnostic test and is not recommended to detect MRD. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.

Note: This Test has been developed and its performance evaluated at Oncquest Laboratories Ltd.

B.Marrow/P Bld. EDTA

MPL Mutation Analysis (Qualitative) ^

PCR & Gene Sequence Analysis

Specimen type: EDTA P Bld

	RESULT _	
	IN RANGE	**OUT OF RANGE**
***MPL* Exon 10 Mutation(s)**	**Not detected**	***

Result:
No mutation was observed in exon 10 of *MPL* gene in the specimen provided.

Regd. Dt:	19/02/2018	Acc. ID: 27178865		Client Details:	Patient Care Centre (Chandigarh)
Coll Dt. Tm:	18/02/2018 00:00:00				SCO No. -140, Sector -240Chandigarh
Recd Dt. Tm:	19/02/2018 10:38:31			Refd. By:	C/O. KOS DIAGNOSTIC LAB
Age:	46 Yrs	Sex:	Male	Report Dt. Tm:	01/03/2018 14:34:56
Name:	Mr. RUPESH GAUR				

Interpretation:

Chronic myeloproliferative disorders (CMPD) are clonal hematopoietic stem cell-disorders characterized by proliferation of one or more myeloid cell lineages. CMPDs include polycythemia vera (PV), essential thrombocythemia (ET), primary myelofibrosis (PMF), chronic myeloid leukemia (CML), hypereosinophilic syndrome (HES), chronic eosinophilic leukemia (CEL) and chronic neutrophilic leukemia (CNL). *JAK2* V617F mutation has been reported in 70-90% cases of PV, 30-70% of ET and 40-50% cases of PMF.

MPL gene found at 1p34 encodes the Thrombopoietin receptor required for platelet production. Mutations at codon 505 and 515 of *MPL* gene have been observed in 3-5% of ET and 8-10% cases of PMF. Identification of *MPL* mutations can aid in the diagnosis of myeloproliferative neoplasms and is highly suggestive of PMF or ET.

Test Attributes and Limitations:

This assay is based upon PCR and Gene Sequencing of Exon 10 of *MPL* (RefSeq NM_005373). The analytical sensitivity of the test allows detection of the mutation when the mutant clone comprises at least 18-20% of the total genomic DNA.

Samples must be received at the laboratory under appropriate conditions within 72hrs of aspiration to ensure preservation of viable high molecular weight DNA. This is a diagnostic test and is not recommended to detect MRD. PCR is a highly sensitive technique; reasons for apparently contradictory results may be due to improper quality control during sample collection, selection of inappropriate specimen and/or presence of PCR inhibitors.

Note: This Test has been developed and its performance evaluated at Oncquest Laboratories Ltd.

***** End of Report *****

Dr. Vinay Bhatia
Ph.D.
Consultant -Molecular Biology

Dr. Sarjana Dutt
Ph.D
Director- Mol.Biology and R&D

The sample is processed by Oncquest Laboratories Ltd.

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