LABORATORY REPORT

Patient Name:	**DUMMY**	**CRM ID:**	NA
Age/DOB:	NA	**Sample Type:**	NA
Sex:	NA	**Collection date:**	DD-MM-YYYY
Referring Clinician:	NA	**Receiving Date:**	DD-MM-YYYY
Test Requested:	**MPL Mutation (Exon10) by Sanger Sequencing**	**Reporting date:**	DD-MM-YYYY

MPL Mutation (Exon10)

CLINICAL DIAGNOSIS/SYMPTOMS

NA

RESULTS

RESULTS	
MPL Mutation (Exon 10)	No variant identified

CLINICALSIGNIFICANCE

Myeloproliferative leukemia gene (MPL) mutations are single base pair substitutions at codons 505 & 515. These mutations have been shown to promote constitutive, cytokine-independent activation of the JAK / STAT signaling pathway and contribute to the oncogenic phenotype. These mutations in exon 10 of MPL have been detected in approximately 5% of patients with Primary myelofibrosis (PMF) and Essential thrombocythemia (ET).

All Lab results are subject to clinical interpretation by qualified medical professional and this report is not subject to use for any medico-legal purpose.

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METHODOLOGY

Targeted sequencing and mutation analysis was performed by Polymerase Chain Reaction (PCR) followed by automated DNA sequencing of the amplicon using BigDye Terminator Chemistry on an ABI Genetic Analyzer 3500XL platform. Sequencing data were aligned to NCBI database to analyze the mutations.

COMMENTS

MPL is located on chromosome 1p34 and encodes the receptor for thrombopoietin, the key growth and survival factor for megakaryocytes. MPL W515L was first described in 2006 amongst JAK2 V617F-negative PMF patients and is the most frequent MPN-associated MPL mutation, resulting from a G to T transition at nucleotide 1544 on exon 10, causing a tryptophan to leucine substitution at codon 515. MPL W515K resulting from TG to AA transition at nucleotide 1543_1544 on exon 10, causing tryptophan to lysine substitution at codon 515. MPL W515A consists of two mutations resulting from TG to GC transition at nucleotide 1543_1544 on exon 10, causing a tryptophan to Alanine substitution at codon 515 resulting from GTG to AGC transition at nucleotide 1542_1544 on exon 10, causing a tryptophan to Alanine substitution at codon 515.

LIMITATIONS

- Indeterminate / Not detected result does not rule out the presence of mutation as it may be below the detection limits of the assay.
- This assay detects W515L, W515K and W515A mutations which form 98% of the reported MPL mutations in Essential thrombocythemia & Primary Myelofibrosis.
- PCR is a highly sensitive technique, however inherent PCR inhibitors in the specimen result in amplification failure.
- The sensitivity of detection for Sanger sequencing is generally recognized as being approximately 15% to 20% mutant allele frequency.

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LABORATORY REPORT

REFERENCES:

1. Jianxiang Chi, Chryso Pierides, Andrie Mitsidou, Andrie Miltiadou, Petroula Gerasimou, Katerina Nicolaou and Paul Costeas. A sensitive detection method for MPLW515L or MPLW515K mutation in myeloproliferative disorders. European Journal of Experimental Biology, 2014, 4(5):33-36

2. Kiladjian JJ, Cassinat B, Turlure P, et al. High molecular response rate of polycythemia vera patients treated with pegylated interferon α-2a. Blood. 2006;108:2037-2040.

3. Pardanani AD, Levine RL, Lasho T, et al. MPL515 mutations in myeloproliferative and other myeloid disorders: astudy of 1182 patients. *Blood.* 2006;108:3472-3476

Conditions of Reporting

- Test results released pertain to the specimen submitted.
- All test results are dependent on the quality of the sample received by the Laboratory.
- Laboratory investigations are only a tool to facilitate arriving at a diagnosis and should be clinically correlated by the Referring Physician.
- Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit the request within 72 hours of postreporting.
- Test results may show inter laboratory variations.
- If Sample collection date is not stated on test requisition form, the current date will be printed by default as the date of collection.
- Test results are not valid for medico legal purposes.

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

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4. This report shall not be deemed valid or admissible for any medico-legal purposes.

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