LABORATORY REPORT

Redcliffe labs

Patient Name	:	Bill Date	:
DOB/Age/Gender	:	Sample Collected	:
Patient ID / UHID	:	Sample Received	:
Referred By	:	Report Date	:
Sample Type	:	Barcode No	:
Client	:	Report Status	:

MOLECULAR DIAGNOSTICS REPORT

BCR ABL1 Kinase Domain Mutation Analysis

Imatinib Resistance Mutation Analysis (Qualitative)

Nested RTPCR and Sanger's Sequencing

Specimen type: EDTA P. Blood

	MUTATION(S) DETECTED	DOMAIN OF MUTATION LOCALIZATION
ABL1 Kinase Domain of BCR/ABL1 transcript	**F 359 V (Phe to Val)**	**Imatinib Binding Domain**

Result:
F359V mutation was detected in the Imatinib Binding Domain of BCR-ABL transcript. This is a clinically relevant mutation and has been previously reported in patients who develop resistance to Imatinib.

The mutant clone comprises 100% of the BCR-ABL transcript load.

Interpretation:
The BCR/ABL1 gene translocation or t(9:22) is found in more than 95% of CML patients, 5% of pediatric ALL-B CALLA positive & 15-30% of adult ALL-B CALLA positive patients. The identification of this translocation stratifies patients that respond to and benefit from Tyrosine Kinase Inhibitor (TKI) therapy. However, a significant minority (5-15%) of patients on treatment may develop secondary resistance to TKIs. The most common mechanism of development of resistance is the emergence of point mutations in the ABL1 kinase domain of the BCR/ABL1 transcript that interfere with Imatinib binding. Patients that exhibit Failure of TKI treatment (as per ELN guidelines) or show an increase of more than 1log in the BCR/ABL1 transcript level should be evaluated for the presence of mutations. Identification of the exact mutation is clinically relevant so that the most effective Second generation TKI may be chosen for further treatment.

Test Attributes and Limitations:
This assay is based upon Nested RTPCR and Gene Sequencing. The analytical sensitivity of the test allows detection of the mutation when the mutant clone comprises at least 18-20% of the total hybrid transcript. Samples must be received at the laboratory under appropriate conditions within 48hrs of aspiration to ensure preservation of viable RNA. 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.

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CONDITIONS OF REPORTING

1. It is Presumed that specimen belongs to patient named or identified, such verification being carried out at the point of generation of said specimen

2. A test might not be performed due to following reason:

- Specimen Quantity not sufficient (Inadequate collection/spillage during transit)
- Specimen Quality not acceptable (Hemolysis/clotted/lipemic.)
- Incorrect sample type
- Test cancelled either on request of patient or doctor

In any of the above case a fresh specimen will be required for testing and reporting

3. The results of the tests may vary from lab to lab ; time to time for the same patient

4. The reported results are dependent on individual assay methods, equipment, method sensitivity, specificity and quality of the specimen received

5. Partial representation of report is not allowed

6. The reported tests are for the notification of the referring doctor, only to assist him/her in the diagnosis and management of the patient

7. If Sample collection date is not stated on test requisition form, the current date will be printed by default as the date of collection.

8. Report with status “Preliminary” means one or more test are yet to be reported

9. This report is not valid for Medico Legal Purpose

10. Applicable Jurisdiction will be of “Delhi” for any dispute/claim concerning the test(s) & results of the test (s)

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

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.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

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

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.