

Case ID 102220373700

Patient Name ARUN DAS

Age/Sex 29 Year /Male

Hospital Location Noida, Uttar Pradesh, India

Hospital Name JITM Skills Private Limited, Noida

Physician Name Dr. Self

Date & Time of Accessioning 19/12/2022 19:00 Hrs

Date & Time of Reporting 22/12/2022 15:13 Hrs

Result



TEST NAME

BCR-ABL1 Kinase Domain Mutations

SPECIMEN INFORMATION

Peripheral Blood Collected on 19/12/2022 at 09:30 Hrs

CLINICAL HISTORY

NOT PROVIDED

METHODOLOGY

Polymerase Chain Reaction

MOLECULAR TEST

RESULT

BCR-ABL1 Kinase Domain Mutations

Negative

INTERPRETATION

No mutation has been detected in the specimen. The result must be correlated carefully with clinical findings. The presence of a kinase domain mutation is critical in treatment plan for the patient. However, other possible factors must also be considered when evaluating treatment response

COMMENTS

- 1. Chronic Myelogenous leukemia (CML) is characterized by the presence of the Philadelphia chromosome (Ph), the product of the t(9;22)(q34;q11) translocation. This translocation results in the BCR-ABL1 fusion protein with constitutive ABL tyrosine kinase activity. Acute myeloid/lymphoid leukemia (AML/ALL) are also known to carry Ph chromosome in a group of patients.
- 2. The first generation tyrosine kinase inhibitor (TKI)- Imatinib is recommended as the first line therapy for BCR-ABL1 positive leukemia that inhibits ABL1 kinase activity.
- 3. A significant number of patients harbor point mutations in the ABL1 kinase domain that renders the leukemic cells resistant to prescribed TKIs.
- 4. This test detects all known as well as novel point mutations in the ABL1 kinase domain that may be leading to therapy resistance.
- 5. Resistance and prognosis is varied for each mutation. Change in dosage, shift to a next-generation TKI are some of the measures to combat therapy resistance.

REFERENCES

- 1. Hochhaus A, Baccarani M, Silver RT, Schiffer C, Apperley JF, Cervantes F, et al. European LeukemiaNet 2020 recommendations for treating chronic myeloid leukemia. Leukemia 2020:1–19. https://doi.org/10.1038/s41375-020-0776-2.
- 2. Radich JP, Deininger M, Abboud CN, Altman JK, Berman E, Bhatia R, et al. Chronic Myeloid Leukemia, Version 1.2019, NCCN





Dr. Shivani Sharma, DCP, DNB

Or. Shivani Sharma, DCP, DNB Reg. No. 1906







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Clinical Practice Guidelines in Oncology. J Natl Compr Canc Netw 2018;16:1108–35. https://doi.org/10.6004/jnccn.2018.0071.

Patient Name

- 3. Hochhaus A, Saussele S, Rosti G, Mahon F-X, Janssen JJWM, Hjorth-Hansen H, et al. Chronic myeloid leukaemia: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol Off J Eur Soc Med Oncol 2018;29:iv261. https://doi.org/10.1093/annonc/mdy159.
- 4.Jabbour E, Kantarjian H. Chronic myeloid leukemia: 2018 update on diagnosis, therapy and monitoring. Am J Hematol 2018;93:442–59. https://doi.org/10.1002/ajh.25011.
- 5. Mughal TI, Radich JP, Deininger MW, Apperley JF, Hughes TP, Harrison CJ, et al. Chronic myeloid leukemia: reminiscences and dreams. Haematologica 2016;101:541–58



Shiwani.

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Question?

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CONDITIONS OF REPORTING

- 1. The tests are carried out in the lab with the presumption that the specimen belongs to the patient named or identified in the bill/test request form.
- 2. The test results relate specifically to the sample received in the lab and are presumed to have been generated and transported per specific instructions given by the physicians/laboratory.
- 3. The reported results are for information and are subject to confirmation and interpretation by the referring doctor.
- 4. Some tests are referred to other laboratories to provide a wider test menu to the customer.
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