# *hema* CŌRE<sup>™</sup> Your Test Results

Case Number:	18014209
Patient Name:	Urmila Devi
Age/Sex:	40 yrs/Female
Patient Location:	Ambala
Hospital Name:	PGI
Physician Name:	Dr. Deepak Goni
Date & Time of Accessioning:	31/03/2018 10:42 Hrs
Date & Time of Reporting:	10/04/2018 17:18 Hrs

#### **TEST NAME**

BCR/Abl Kinase Domain Mutations (IRMA)

#### **SPECIMEN INFORMATION**

Received peripheral blood in EDTA collected on 29/03/2018.

**CLINICAL HISTORY** 

#### Not Provided

#### **METHODOLOGY**

Polymerase Chain Reaction - Sequencing.

**MOLECULAR TEST** 

**BCR Abl Kinase Domain Mutations** 

Tested

T315I

E255V

Y253H

E255K

Y253F

E373G

L248V

G321E

E279K

G250E

D276G

F486S

#### BCR/Abl Kinase Domain Mutations (IRMA)

Results	BCR Abl Kinase Domain Mutations Tested	Results	l Do
Wild Type	H396P	Wild Type	V3
Wild Type	H396R	Wild Type	E35
Wild Type	F359V	Wild Type	M3
Wild Type	F317L	Wild Type	T31
Wild Type	Q252H	Wild Type	M2
Wild Type	L387M	Wild Type	F31
Wild Type	E275K	Wild Type	L38
Wild Type	F359C	Wild Type	V29
Wild Type	E450K	Wild Type	F31
Wild Type	M351T	Wild Type	G2
Wild Type	M244V	Wild Type	V28
Wild Type	E355G	Wild Type	Y23

#### INTERPRETATION

#### Negative

BCR Abl Kinase Domain Mutations Tested	Results	
V379I	Wild Type	
E355A	Wild Type	
M388L	Wild Type	
T315A	Wild Type	
M237I	Wild Type	
F311L	Wild Type	
L387F	Wild Type	
V299L	Wild Type	
F317V	Wild Type	
G250A	Wild Type	
V289A	Wild Type	
Y235H	Wild Type	

Dr. Rahul Katara, Ph.D., Molecular Scientist



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#### COMMENTS

- 1. Chronic Myelogenous leukemia (CML) is characterized by the presence of the Philadelphia chromosome, the product of the t(9;22)(q34;q11) translocation. This translocation results in the BCR/ABL fusion protein with constitutive ABL tyrosine kinase activity.
- 2. The kinase inhibitor imatinib (STI571, Gleevac) inhibits ABL kinase activity and is now standard of care for early phase CML. Prolonged treatment with Imatinib can lead to drug resistance, especially in patients with advanced disease.
- A large portion of resistant patients have acquired point mutations in the ABL kinase domain that renders the kinase, resistant to 3. the drug. Site of point mutations in ABL associated with Imatinib resistance span the entire kinase domain but often cluster in important hotspots.
- This test detects greater that 90 percent of ABL mutations that may lead to Imatinib resistance, including the important T3151 4. and P-loop mutations. A range of levels of resistance and prognosis has been observed for different mutations. ABL kinase domain mutations that cause only moderate resistance may be overcome by higher Imatinib doses.

#### Assay Description And Methodology:

Total RNA extracted via common sample preparation methodologies from whole blood or bone marrow collected in EDTA is compatible with PCR Sequencing methods. The PCR product sequence is aligned with the Wild Type sequence to detect the codons (HOT SPOT Region) of interest.

#### Intended Use:

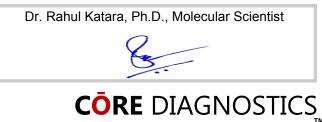
The BCR-ABL Kinase domain mutation Detection Kit is intended for the qualitative detection of BCR-ABL KD mutation in bone marrow or peripheral blood samples using conventional PCR system. The kit is based on reverse transcription of total RNA, followed by Sequencing based PCR amplification and detection of BCR-ABL KD mutation by sanger sequencing.

#### **Disclaimer:**

This test is performed using in-house developed primers for BCR ABL & KD mutation. The assay is designed to perform the reactions at the specified analytical sensitivity given that the template RNA is not heavily fragmented and does not contain materials that could inhibit the amplification reaction.

#### REFERENCES

- 1. BaccaraniM, Saglio G, Goldman J, Hochhaus A, Simonsson B, Appelbaum F, Apperley J, Cervantes F, Cortes J, Deininger M, Gratwohl A, Guilhot F, Horowitz M, Hughes T, Kantarjian H, Larson R, Niederwieser D, Silver R, Hehlmann R; European LeukemiaNet. Evolving concepts in the management of chronic myeloid leukemia: recommendations from an expert panel on behalf of the European LeukemiaNet. Blood 2006Sep 15;108(6):1809-20.
- 2. Beillardet al. Evaluation of candidate control genes for diagnosis and residual disease detection in leukemic patients using 'real-time' quantitative reverse-transcriptase polymerase chain reaction (RQ-PCR) – a Europe against cancer program. Leukemia 200317, 2474 - 2486.
- 3. BranfordS, Cross NC, Hochhaus A, Radich J, Saglio G, Kaeda J, Goldman J, Hughes T. Rationale for the recommendations for harmonizing current methodology for detecting BCR-ABL transcripts in patients with chronic myeloid leukaemia. Leukemia 2006Nov;20(11):1925-30.
- 4. DrukerBJ, Guilhot F, O'Brien SG, Gathmann I, Kantarjian H, Gattermann N, Deininger MW, Silver RT, Goldman JM, Stone RM, Cervantes F, Hochhaus A, Powell BL, Gabrilove JL, Rousselot P, Reiffers J, Cornelissen JJ, Hughes T, Agis H, Fischer T, Verhoef G, Shepherd J, Saglio G, Gratwohl A, Nielsen JL, Radich JP, Simonsson B, Taylor K, Baccarani M, So C, Letvak L, Larson RA; IRIS Investigators. Fiveyear follow-up of patients receiving imatinib for chronic myeloid leukemia. N Engl J Med. 2006Dec 7;355 (23):2408-17.



### **Questions?**

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