CŌRE DIAGNOSTICS™

Your Test Results

Case Number:

Patient Name:

Age/Sex:

Patient Location:

Hospital Name:

Physician Name:

Date & Time of Accessioning:

Date & Time of Reporting:

TEST NAME

MYD88, Mutation Analysis

SPECIMEN INFORMATION

Received peripheral blood in EDTA, collected on 29/09/2023 at 11:00 Hrs

CLINICAL HISTORY

Not Provided

METHODOLOGY

Sanger Sequencing

RESULTS

Gene	Variant tested	Remark
MYD88	c.794T>C;p.L265P	**Not Detected**

*The results should be considered in conjunction with clinical information, histologic evaluation, and/or additional testing.

COMMENTS

1. This assay will only detect the above mentioned targeted mutations.
2. Any other mutations present within the exons, promoter, the 5' and 3' untranslated regions, and regions deeper (than 10-20 nucleotides) within the introns will not be detected.
3. Large deletions that eliminate most or all of the coding sequence will also not detected by this assay.
4. The result does not rule out the presence of a mutation that may be present below the limit of detection for this assay (20%).
5. False-negative results may occur in specimens when tumor cells comprise <40% of the cell population.
6. Rare diagnostic errors can occur due to probe-site mutations.

CLINICAL BACKGROUND

1. This assay detects mutations in the MYD88 gene, which encodes a cytosolic adapter protein that plays a central role in the innate and adaptive immune response.
2. This protein functions as an essential signal transducer in the interleukin-1 and Toll-like receptor signaling pathways. These pathways regulate that activation of numerous proinflammatory genes.
3. The encoded protein consists of an N-terminal death domain and a C-terminal Toll-interleukin1 receptor domain.
4. Patients with defects in this gene have an increased susceptibility to pyogenic bacterial infections.
5. Alternate splicing results in multiple transcript variants.Diseases associated with MYD88 include Myd88 Deficiency and Waldenstrom Macroglobulinemia.

Dr. Sanjay Kumar, Ph.D.

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

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CLINICAL BACKGROUND

6. The MYD88 L265P mutation results in an amino acid substitution at position 265 in exon 5, from a leucine (L) to a proline (P).
7. This mutation occurs within the Toll-IL1 receptor domain of the gene and is predominantly seen in patients diagnosed with Waldenstrom Macroglobulinemia (WM).
8. MYD88 L265P is a gain of function mutation which results in increased cell survival by increasing NF-κB activity, JAK-STAT3 signaling, and consequently cytokine production.
9. The presence of L265P mutation confers a higher probability of disease progression and a poor response to therapies, making it an adverse prognostic factor as compared to wild-type gene.

REFERENCES

1. Lord KA, Hoffman-Liebermann B, Liebermann DA (Jul 1990). "Nucleotide sequence and expression of a cDNA encoding MyD88, a novel myeloid differentiation primary response gene induced by IL6". Oncogene. 5 (7): 1095–7.
2. Treon SP, Xu L, Yang G, Zhou Y, Liu X, Cao Y, Sheehy P, Manning RJ, Patterson CJ, Tripsas C, Arcaini L, Pinkus GS, Rodig SJ, Sohani AR, Harris NL, Laramie JM, Skifter DA, Lincoln SE, Hunter ZR (2012). "MYD88 L265P somatic mutation in Waldenström's macroglobulinemia". N. Engl. J. Med. 367 (9): 826–33.

Dr. Sanjay Kumar, Ph.D.

Dr. Shivani Sharma, DCP, DNB
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Question?

Contact us at **+91 124 4615 615**

Toll Free Helpline **+91 8882899999**

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.
5. Core Diagnostics Pvt. Ltd. shall in no event be liable for accidental damage, loss, or destruction of specimen, which is not attributable to any direct and mala fide act or omission of Core Diagnostics Pvt. Ltd. or its employees. Liability of Core Diagnostics Pvt. Ltd. for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.

CORE Diagnostics National Reference Lab - Gurugram (102)

406, Udyog Vihar, Phase III, Gurgaon 122016

CORE Diagnostics Lab - New Delhi (103)

C-13, Green Park Extension, New Delhi – 110016

CORE Diagnostics Lab - Bangalore (105)

1st Floor, KMK Tower, 142 KH Road, Bangalore - 560027

CORE Diagnostics Lab - Lucknow (109)

J.S. Tower, Plot No. K-702, Sector-K, Ashiyana,

Near Raj Luxmi Sweets, Lucknow-226012

CORE Diagnostics Lab - Bhubaneswar (108)

Plot No. 249, Near Police Academy, AIIMS Nagar,

Patrapada, Bhubaneswar-751019

This is a transcribed report and the test was performed at the laboratory OSL 14