IgVH/IgHV Mutation Analysis

PATIENT	REPORT DATE	BOOKING ID
Bhanu P Singh	21 March 2022	#012202260070

Test Description

This test is designed to detect mutation status of the immunoglobulin heavy chain variable region gene in clonal B cell populations. A clonal rearrangement was detected utilizing VH gene segment using PCR sequencing.

This assay is useful to provide an estimate of prognosis (aggressiveness of disease) of B-cell chronic lymphocytic leukemia (B-CLL) and to help evaluate treatment options B-CLL with mutated IgVH is typically less aggressive (more slowly progressive) than B-CLL with unmutated IgVH

Patient Demographic

Name: Mr Bhanu Pratap Singh
Sex: Male
Date of Birth/Age: 50 years
Disease: Chronic Lymphocytic Leukemia (CLL)

Clinician

Clinician Name: Dr Amit Verma
Medical Facility: Dr AV Institute of Personalized Therapy and Cancer Research (IPTCR)
Pathologist: Not Provided

Specimen

Booking ID: 012202260070
Sample Type: Peripheral Blood
Date of Collection: 26-02-2022
Date of Booking: 26-02-2022

RESULTS

IgVH gene mutation was detected

INTERPRETATION

1. IGVH Hypermutation status is analyzed using the immunoglobulin databases available on line International Immunogenetics Information (IMGT) (http://imgt.cines.fr) – IMGT V-Quest and GenBank (http://www.ncbi.nlm.nih.gov/igblast/).
2. The results are reported as percentage of homology between the patient's VH sequence in comparison with the germ-line VH sequence.
3. The patient is considered positive for somatic hyper-mutation when the mutation rate is >2.0%.

COMMENTS

1. B-cell chronic lymphocytic leukemia {CLL) is the most frequent form of leukemia in adults which is characterized by a progressive accumulation of functionally incompetent B-cells.
2. The disease is a highly variable in clinical course. Some patients with CLL survive for decades and never require therapy others succumb rapidly to the disease despite therapy.
3. Those patients showing unmutated IGVH (<2% somatic mutation) have a relatively aggressive disease course with an average survival of 79- 117 months. Those patients with hypermutated IGVH (>2% somatic mutation) have a longer average survival of 124-293 months. The assay detects somatic mutations in the variable region of the immunoglobulin heavy chain gene locus (lgVH) by PCR-Sequencing.

Jatinder Kaur, PhD
Head, Molecular Biology & Genomics

Dr. Gulshan Yadav, MD
Head, Pathology

LIMITATIONS

- The sensitivity of the assay is 10%. The assay cannot be performed on specimen containing less than 10% clonal B-cells.
- PCR is a highly sensitive technique however inherent PCR inhibitors in the specimen may result into amplification failure.
- Results of this test must always be interpreted in the context of clinicopathologic findings, family history and other relevant clinical and laboratory data.

REFERENCES

1. Bilous N, *et al.* Significance of VH genes mutation status for prognosis of CLL patients. Exp Oncol. 2005 Dec; 27(4): 325-9.
2. Mutated or non-mutated? Which database to choose when determining the IgVH hypermutation status in chronic lymphocytic leukemia? Haematologica 2006; 91(1): 11-12.
3. Ghia P, *et al.* ERIC recommendations on IGHV gene mutational status analysis in chronic lymphocytic leukemia. Leukemia. 2007 Jan; 21(1): 1-3.

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. MolQ Laboratory 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 MolQ Laboratory or its employees. Liability of MolQ Laboratory for deficiency of services, or other errors and omissions shall be limited to fee paid by the patient for the relevant laboratory services.

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