Brief Report
Volume 13:5, 2022

Journal of Molecular Biomarkers & Diagnosis

ISSN: 2155-9929 Open Access

Brief Report on Genomic Analysis Using Cell-free DNA in Mediastinal B-cell Lymphoma

Marlon Oliveira*

Department of Molecular Biology, University of Cairo, Cairo, Egypt

About the Study

The World Health Organization (WHO) classifies primary mediastinal large B-cell lymphoma (PMBL) as a distinct entity with distinct clinical, histological, and molecular characteristics. It is responsible for 2% to 3% of all non-Hodgkin lymphomas. Patients typically present with a large mass in the anterior mediastinum, which makes biopsy difficult. Histological samples can occasionally be non-diagnostic due to extensive fibrosis and necrosis, requiring patients to undergo mediastinoscopy or thoracoscopy to determine the diagnosis. PMBL is characterized by recurrent genomic alterations such as somatic gene mutations and copy number alterations (CNA), as well as a distinct gene expression profile.

The disease is characterized by constitutive activation of the nuclear factor-kB (NF-kB) and JAK/STAT pathways. Cell-free DNA (cfDNA) has emerged as a noninvasive tool to supplement tissue biopsies, particularly in cases where obtaining a tumour biopsy is clinically difficult. In oncology, cfDNA has proven useful in monitoring treatment response in real time, guiding therapy, and detecting early recurrence. In recent years, cfDNA has been studied in Hodgkin and non-Hodgkin lymphomas using next-generation sequencing (NGS) for genetic analysis, providing a simple and easy detection method for assisting in tumour molecular profiling [1-3].

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Future Perspective

Several studies of cfDNA in diffuse large B-cell lymphoma (DLBCL) using NGS customized gene panels found that the mutational landscape from cfDNA samples was highly consistent with that observed in tissue biopsies, and also highlighted mutations found only in cfDNA, which could be explained by the tumor's spatial heterogeneity. Despite the fact that some of these studies include PMBL, the small number of cases studied and the lack of PMBL-specific analyses preclude any firm conclusions about the use of cfDNA for PMBL genomic characterization. Genetic studies in PMBL are difficult due to the scarcity of available material. This is especially important during diagnosis, when a large core needle is used to obtain a biopsy that yields enough material to establish a diagnosis but not enough for further analysis, and during relapse. In this context, the use of cfDNA may overcome this limitation and serve as a reliable foundation for genetic research in PMBL.

Thus, the goal of this study was to evaluate the use of cfDNA as a reliable source for genomic characterization in newly diagnosed patients with PMBL in a real-world setting, using a custom NGS panel for gene mutations and low-pass whole-genome sequencing (WGS) for CNA, and its correlation with clinical parameters. cfDNA analysis is increasingly being used in oncology, including patients with lymphoma, to assess molecular profiling at diagnosis, define prognosis, and identify therapeutic targets . We investigated the suitability of cfDNA as a reliable source for mutational and CNA assessment in PMBL patients, where diagnostic biopsies are frequently difficult to obtain. Indeed, information on the utility of cfDNA in PMBL is limited, with only one recent report. We were able to detect mutations in the cfDNA in 18 of 20 (90%) cases in the current series, a rate comparable to that reported in other lymphoma series, including a DLBCL cohort from our own institution.

The mutational profile of 44 patients with PMBL was reported using an abridged targeted panel of nine genes, with at least one mutation detected in 32 (73 percent) of patients. The higher detection rate in our study could be explained by the fact that we used a broader gene panel, including CNA analysis, and that we used molecular-barcode technology to improve background removal in sequencing data analysis and increase sensitivity. PMBL has been associated with a variety of genetic alterations, including constitutive activation of the NF-B and JAK-STAT pathways, as well as genetic alterations that promote immune evasion. The mutational landscape described in our study from cfDNA was highly consistent with that previously published in various series, including two integrative genetic analyses. SOCS1, STAT6, NFKBIA, NFKBIE, and TNFAIP3 were among the most frequently altered genes, as were members of the JAK-STAT [4,5] and NF-kB pathways. Gains of 9p (CD274 and PDCD1LG2), 2p (REL), chromosome 6, and 11q have all been described in PMBL.

Conflict of Interest

None.

Acknowledgement

None.

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***Address for Correspondence:** Marlon Oliveira, Department of Molecular Biology, University of Cairo, Cairo, Egypt, E-mail: MarlonOliveira5@gmail.com*

Received: 02-May-2022, Manuscript No. jmbd-22-68161; **Editor assigned:** 04-May-2022, Pre QC No. P- 68161; **Reviewed:** 18-May-2022, QC No.Q-68161; **Revised:** 23-May-2022, Manuscript No.R-68161; **Published:** 30-May-2022, DOI:10.37421/2155-9929.2022.13.525

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How to cite this article: Oliveira, Marlon. “Brief Report on Genomic Analysis Using Cell-free DNA in Mediastinal B-cell Lymphoma.” J Mol Biomark Diagn 13 (2022): 525.