

LIQUID BIOPSY-BASED DETECTION AND MONITORING OF EZH2 GENE MUTATIONS IN FOLLICULAR LYMPHOMA

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Laura Kiss was born and raised in Kiskunfelegyháza, Hungary. Currently she is a final year medical student at Semmelweis University in Budapest. She is an active member of the scientific student research community of the university; her research project focuses on molecular oncohematology.

Aim: Follicular lymphoma (FL) has an overall survival rate of 15 years, however the disease is incurable with current standard of care. The majority of patients experience disease progression, raising the need for effective treatment response monitoring. Molecular analysis of tumor derived circulating DNA (ctDNA), referred as liquid biopsy, is a promising minimally invasive disease monitoring tool for patients suffering from broad spectrum of tumor types. Recent molecular profiling studies have identified gain of function mutations in 25% of FL cases in the EZH2 gene. The aim of our research was to develop a liquid biopsy-based EZH2 mutation test for non-invasive testing.

Methods: Pretreatment plasma samples were collected from 107 patients with active FL. To characterize the EZH2 mutation status, samples were tested by digital droplet PCR method (Bio-Rad, USA). In a multiplex PCR setup, we analyzed seven point mutations performing only two reactions.

Results: EZH2 mutations were detected in 29% (31/107) of the patients, while 71% of the cases proved to be wild-type. In EZH2 positive patients correlation between variant allele frequency (VAF) and therapy efficacy was demonstrated by analyzing samples collected during treatment. 78% (18/23) of evaluable patients responded to treatment, meanwhile 22% were refractory to therapy.

Conclusion: Our results suggest that liquid biopsy-based EZH2 mutation analysis is applicable for minimal invasive detection and monitoring of EZH2 mutations in FL. The frequency of EZH2 mutations was found to be 29%, somewhat higher than previously found in tissue samples. Our observation raises the possibility of a higher incidence of EZH2 mutations in FL, which may further expand the set of patients who may be eligible for EZH2 inhibitor therapy in the future.



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53rd Annual Scientific Meeting of HMAA, Oct. 23 – Oct. 28, 2022

Certificate of attendance

Date: October 28, 2022

To Whom It May Concern:

Laura Kiss attended our 53rd Annual HMAA scientific symposium in Sarasota, Florida, between October 23 – October 28, 2022.

Best regards,

Istvan Stadler Ph.D.

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