Preliminary European resultson the clinicalutility of ColonAiQ, a cell-free DNA methylation-based liquid biopsy assay for colorectal cancer early detection

Dávid Kis[1], Péter Lukász[2], Anikó Serdült[1], Abigél Balla[1,3], Péter Hunyadi[1], Balázs Bánky[2], Attila Szijártó[2], Orsolya Biró[1,3]

[1]Clinomics Europe Ltd.
[2]Semmelweis University, Faculty of Medicine, Department of Surgery, Transplantation and Gastroenterology
[3]Semmelweis University DoctoralSchool, SurgicalMedicineDivision

Background: Colorectal cancer (CRC) is one of the most common and deadliest cancers worldwide. Early detection can significantly improve patient survival. In this report, we present the preliminary European results of ColonAiQ, a multi-gene methylation liquid biopsy assay that can detect pre-cancerous lesions and early-stage colorectal cancer.

Methods: Study participants were recruited at Semmelweis University in 2023. The distribution of the patient group classified histopathologically was as follows: 16.17 % (4/24) had non-neoplastic gastrointestinal disorders (GI), 20.83 % (5/24) had adenomas and polyps(A), 16.67 % (4/24) had CRC I-II stages, and 55.83 % (11/24) had CRC III stage. The study included 23 healthy controls matched for age and gender. Peripheral blood was collected in EDTA tubes one day before surgery. Cell-free DNA (cfDNA) was extracted from 3-4 mL of non-hemolysed plasma and analyzed using multiplex methylation-specific qPCR (ColonAiQ). The detection rate was compared to that of the standard tumor markers (CEA and CA19-9).

Results: All cfDNA samples passed quality and quantity criteria. CEA/CA19-9 levels were elevated in 50/25 % of GI group, 0/0 % of A group, 0/0 % of CRC I-II group, and 36.36/18.18 % of CRC III group. ColonAiQ-positive samples were found in 25 % of GI, 40 % of A, 75 % of CRC I-II, and 81.82 % of CRC III patients. The co-methylation of 4 to 5 regions of Septin9-1, Septin9-2, BCAT1, IKZF1, and VAV3 was limited to advanced CRC samples, while single or several co-methylated regions were found in polypous, adenomatous, and various CRC patients. The healthy control group had a positive rate of 4.35 % (1/23) for the Septin9-2 marker.

Conclusion: We demonstrated the clinical utility of ColonAiQ, a blood-based assay for detecting colorectal cancer (CRC) in a European subpopulation. The ColonAiQ assay is a liquid biopsy workflow that can be easily implemented in the clinic, potentially reducing the morbidity and mortality of CRC. Further investigations are planned on a larger patient cohort.