Colon AiQ

SAMPLE INFORMATION

Name		**Date Received**	
Medical ID		**Date Of Report**	
Material		**Barcode**	

RESULTS

Positive: An abnormal methylation signal was detected

- This signal in free circulating DNA (cfDNA) has been associated with the presence of malignancy in the colon.
- It could also be related to other abnormal conditions of the colon, including adenomas and colon polyps.
- A positive result should not be used for colon cancer diagnosis. A colonoscopy is required to confirm the diagnosis.

RECOMMENDATIONS

Medical screening and management should rely on clinical findings and family history. Follow-up examinations, such as colonoscopy and histopathologic examination, are recommended to detect any possible precancerous or cancerous lesions in this patient. It is recommended to evaluate the result with the attending physician.

GENOMIX

Leoforos Marathonos 46, 15351, Pallini, Greece

2160005494

www.iqgenomix.com

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METHODOLOGY

Free circulating DNA was isolated from the examined sample (QIAamp Circulating Nucleic Acid Kit, Qiagen). DNA modification was performed with sodium bisulfite. This was followed by methylation analysis in the promoter of *Septin9*, *IKZF1, BCAT1* and *VAV3* genes by preamplification and fluorescent quantitative PCR, using the ColonAiQ CE-IVD test.

INFORMATION ABOUT THE TEST

Colorectal cancer (CRC) is one of the most common and deadly cancers worldwide. 5-year survival for CRC ranges from 91% (localized) when detected early to 13% (distant) when diagnosed late (SEER database). American Cancer Society recommends people with average CRC risk to start regular screening at age 45.

Circulating cell-free DNA (cfDNA) in biological fluids such as blood (plasma/serum) contains circulating tumor DNA (ctDNA) which shows epigenetic alterations associated with cancer development. Several studies have demonstrated that DNA-methylation profiling in cfDNA isolated from blood plasma can be effectively utilized in early cancer detection καρκίνου (PMID: 32694610, 34176681). Using the methylation signature of ctDNA released from colorectal cancer, this CRC early detection assay showed a sensitivity of 86% and a specificity of 92% in a published study including 173 CRC patients (Stages I to IV), 107 patients with advanced adenomas (AA), and 136 colonoscopy-negative controls (PMID: 34487783).

Utilization of this assay in CRC patients pre- and post-therapy has also been shown to allow assessment of recurrence risk and to enable early detection of recurrence (PMID: 37079312).

LIMITATIONS OF THE TEST

- 50% of positive patients will have a positive colonoscopy. A positive signal cannot be used to diagnose colon cancer without positive findings on colonoscopy. Some patients without colon cancer may have a detectable signal.
- The test cannot detect all patients with colon cancer.
- A negative result cannot be used to rule out a diagnosis of colon cancer.
- The test does not replace other colon cancer screening tests recommended by a healthcare provider.
- Each molecular analysis has an internal error probability of 0.5-1%. This is due to rare molecular events and factors related to sample preparation and analysis.

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REFERENCES

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