

Detection rate for *esr1* mutations is higher in circulating-tumor-cell-derived genomic dna than in paired plasma cell-free dna samples as revealed by ddpcr

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Abstract

Plasma cell-free DNA (cfDNA) analysis to track estrogen receptor 1 (ESR1) mutations is highly beneficial for the identification of tumor molecular dynamics and the improvement of personalized treatments for patients with metastatic breast cancer (MBC). Plasma-cfDNA is, up to now, the most frequent liquid biopsy analyte used to evaluate ESR1 mutational status. Circulating tumor cell (CTC) enumeration and molecular characterization analysis provides important clinical information in patients with MBC. In this study, we investigated whether analysis of CTCs and circulating tumor DNA (ctDNA) provide similar or complementary information for the analysis of ESR1 mutations. We analyzed both plasma-cfDNA (n = 90) and paired CTC-derived genomic DNA (gDNA; n = 42) from 90 MBC patients for seven ESR1 mutations using the ddPLEX Mutation Detection Assay (Bio-Rad, Hercules, CA, USA). Eight out of 90 (8.9%) plasma-cfDNA samples tested with ddPCR were found positive for one ESR1 mutation, whereas 11/42 (26.2%) CTC-derived gDNA samples were found positive for at least one ESR1 mutation. Direct comparison of paired samples (n = 42) revealed that the ESR1 mutation rate was higher in CTC-derived gDNA (11/42, 26.2%) than in plasma-cfDNA (6/42, 14.3%) samples. Our results, using this highly sensitive ddPLEX assay, reveal a higher percentage of mutations in CTC-derived gDNAs than in paired ctDNA in patients with MBC. CTC-derived gDNA analysis should be further evaluated as an important and complementary tool to ctDNA for identifying patients with ESR1 mutations and for guiding individualized therapy.

Do you have any conflicts of interest?

No, I do not have a conflict of interest.