

Next-Generation Sequencing of Circulating Tumour DNA for Early Detection

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ABSTRACT

Circulating tumour DNA enables non-invasive cancer detection. We developed a next-generation sequencing assay targeting methylation and mutation signals simultaneously, validated in a prospective cohort of 1,150 participants. The combined assay achieved 84% sensitivity at 99.4% specificity for stage I-II disease, substantially exceeding mutation-only detection. Tissue-of-origin prediction was correct in 88% of positive cases.

KEYWORDS

circulating tumour DNA, liquid biopsy, early detection, next-generation sequencing

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