METHOD FOR DETECTION AND QUANTIFICATION OF GENETIC ALTERATIONS

ABSTRACT

Disclosed is a method of simultaneously capturing and identifying a defined target region and a partly defined target region within a DNA sample, wherein the partly defined target region comprises a structural variation or rearrangement or fusion. Firstly, a barcoded primer will bind to fragments comprising the defined target region, and another barcoded primer comprising a separation molecule will bind to fragments comprising the partly defined target region. Secondly, the primers that annealed to the defined target region (i.e. product A) are separated from the primers that annealed to the partly defined target region (i.e. product B). Thirdly, the two products are treated differently. For product A, a reverse primer will be added. For product B, a double stranded oligonucleotide is ligated to the end that is not connected to the separation molecule. Fourthly, product A and product B that has been processed are recombined, amplified together, and the resulting amplicons are sequenced.

Fig. 1B