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upQMPSF, a Method for the Detection of BRCA1 Exon Copy Number Variants

??? ??????: 08 ????/???? 2017

Abstract

Large insertions/deletions mutations are frequently found in genes associated with certain diseases such as hereditary cancers.

These mutations are mostly overlooked by current classical screening techniques due to their certain limitations. This justifies the need to improve the existing techniques or design novel ones.

A modified version of quantitative multiplex PCR short fluorescent fragment (QMPSF), termed universally primed QMPSF (upQMPSF), was developed.

The modifications enhance multiplexing capacity, reduce cost, and improve the mutation detection spectrum.

upQMPSF was used to screen germline mutations in 88 familial ovarian cancer patients negative for point mutations. upQMPSF successfully detected a 2.8 kb copy number gain spanning exon 15 of BRCA1 gene mediated by Alu-Alu homologous-based recombination.

upQMPSF is a cost-efficient, versatile method, and demonstrated efficiency in detecting structural variations as a potential method for genetic testing in clinical and .research laboratories

25991562PMID :