Title (en)

METHOD FOR DETECTION AND QUANTIFICATION OF GENETIC ALTERATIONS

Title (de)

VERFAHREN ZUR DETEKTION UND QUANTIFIZIERUNG VON GENETISCHEN VERÄNDERUNGEN

Title (fr)

PROCÉDÉ DE DÉTECTION ET DE QUANTIFICATION DE MODIFICATIONS GÉNÉTIQUES

Publication

EP 3810805 A4 20220323 (EN)

Application

EP 19825310 A 20190625

Priority

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- SG 2019050317 W 20190625

Abstract (en)

[origin: WO2020005159A1] 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.

IPC 8 full level

C12Q 1/6855 (2018.01); **C12Q 1/6858** (2018.01); **C12Q 1/6869** (2018.01)

CPC (source: EP)

C12Q 1/6855 (2013.01); **C12Q 1/6858** (2013.01); C12N 9/1252 (2013.01)

Citation (search report)

- [Y] WO 2015039006 A1 20150319 - GEN HOSPITAL CORP [US]
- [Y] WO 2018111835 A1 20180621 - DANA FARBER CANCER INST INC [US]
- [Y] WO 2014093330 A1 20140619 - CLEARFORK BIOSCIENCE INC [US]
- [Y] WO 2011019964 A1 20110217 - NUGEN TECHNOLOGIES INC [US], et al
- [Y] SCOTT R KENNEDY ET AL: "Detecting ultralow-frequency mutations by Duplex Sequencing", NATURE PROTOCOLS, vol. 9, no. 11, 9 October 2014 (2014-10-09), GB, pages 2586 - 2606, XP055745195, ISSN: 1754-2189, DOI: 10.1038/nprot.2014.170
- [Y] QING WANG ET AL: "Targeted sequencing of both DNA strands barcoded and captured individually by RNA probes to identify genome-wide ultra-rare mutations", SCIENTIFIC REPORTS, vol. 7, no. 1, 13 June 2017 (2017-06-13), XP055551336, DOI: 10.1038/s41598-017-03448-8
- See references of WO 2020005159A1

Designated contracting state (EPC)

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