Title (en)

METHODS FOR DETERMINING ABSOLUTE GENOME-WIDE COPY NUMBER VARIATIONS OF COMPLEX TUMORS

Title (de)

VERFAHREN ZUR BESTIMMUNG DER ABSOLUTEN GENOMWEITEN KOPIENZAHLVERÄNDERUNGEN KOMPLEXER TUMOREN

Title (fr)

PROCÉDÉS DE DÉTERMINATION DES VARIATIONS DU NOMBRE DE COPIES ABSOLU À L'ÉCHELLE DU GÉNOME DE TUMEURS COMPLEXES

Publication

EP 2844771 A4 20151202 (EN)

Application

EP 13784660 A 20130506

Priority

- US 201261643225 P 20120504
- US 2013039777 W 20130506

Abstract (en)

[origin: WO2013166517A1] Methods for interpreting absolute copy number of complex tumors and for determining the copy number of a genomic region at a detection position of a target sequence in a sample are disclosed. In certain aspects, genomic regions of a target sequence in a sample are sequenced and measurement data for sequence coverage is obtained. Sequence coverage bias is corrected and may be normalized against a baseline sample. Hidden Markov Model (HMM) segmentation, scoring, and output are performed, and in some embodiments population-based no-calling and identification of low-confidence regions may also be performed. A total copy number value and region-specific copy number value for a plurality of regions are then estimated.

IPC 8 full level

C12Q 1/68 (2006.01); **G16B 20/10** (2019.01); **G16B 20/20** (2019.01); **G16B 30/10** (2019.01); **G16B 30/20** (2019.01)

CPC (source: CN EP US)

C12Q 1/6827 (2013.01 - CN EP); **C12Q 1/6886** (2013.01 - CN EP); **G16B 20/00** (2019.01 - EP); **G16B 20/10** (2019.01 - CN EP US); **G16B 20/20** (2019.01 - CN EP US); **G16B 30/00** (2019.01 - CN EP); **G16B 30/10** (2019.01 - CN EP US); **G16B 30/20** (2019.01 - CN EP US); C12Q 2600/156 (2013.01 - CN EP)

Citation (search report)

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Designated contracting state (EPC)

AL AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HR HU IE IS IT LI LT LU LV MC MK MT NL NO PL PT RO RS SE SI SK SM TR

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DOCDB simple family (application)

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