

Title (en)

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

Title (de)

VERFAHREN ZUR BESTIMMUNG DER ABSOLUTEN GENOMWEITEN KOPIENZAHIVERÄNDERUNGEN 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

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- 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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