

Title (en)
METHODS AND SYSTEMS FOR A LIQUID BIOPSY ASSAY

Title (de)
VERFAHREN UND SYSTEME FÜR EINEN FLÜSSIGBIOPSIETEST

Title (fr)
PROCÉDÉS ET SYSTÈMES DE DOSAGE DE BIOPSIE DE LIQUIDE

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EP 4107732 A1 20221228 (EN)

Application
EP 21711691 A 20210218

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- US 202063041293 P 20200619
- US 202063041272 P 20200619
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Abstract (en)
[origin: WO2021168146A1] Methods, systems, and software are provided for validating a copy number variation, validating a somatic sequence variant, and/or determining circulating tumor fraction estimates using on-target and off-target sequence reads in a test subject. A copy number status annotation for a genomic segment is validated by applying a first dataset to a plurality of filters comprising a measure of central tendency bin-level sequence ratio filter, a confidence filter, and a measure of central tendency-plus-deviation bin-level sequence ratio filter. A somatic sequence variant is validated by comparing a variant allele fragment count for a candidate somatic sequence variant for a respective locus, against a dynamic variant count threshold for the locus in a respective reference sequence. A circulating tumor fraction is estimated based on a measure of fit between genomic segment-level coverage ratios and integer copy states across a plurality of simulated circulated tumor fractions.

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