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(54) Title (EN): TARGET-ENRICHED MULTIPLEXED PARALLEL ANALYSIS FOR ASSESSMENT OF TUMOR BIOMARKERS

(54) Title (FR): ANALYSE PARALLÈLE MULTIPLEXÉE ENRICHIE EN CIBLE POUR L'ÉVALUATION DE BIOMARQUEURS TUMORAUX

(57) Abstract:

(EN): The invention provides methods for assessment of tumor biomarkers in biological samples using target-enriched multiplexed parallel analysis. The methods of the invention utilize TArget Capture Sequences (TACS) to thereby enrich for target sequences of interest, followed by massive parallel sequencing and statistical analysis of the enriched population. The methods can be used with DNA samples from a patient, such as a tissue biopsy or plasma sample (liquid biopsy), for detection of the presence of tumor biomarkers, e.g., for purposes of diagnosis, screening, therapy selection and/or treatment monitoring. Kits for carrying out the methods of the invention are also provided.

(FR): L'invention concerne des méthodes d'évaluation de biomarqueurs tumoraux dans des échantillons biologiques à l'aide d'une analyse parallèle multiplexée enrichie en cible. Les méthodes de l'invention utilisent des Séquences de Capture Cibles (TACS) pour enrichir ainsi des séquences cibles d'intérêt, suivi par un séquençage parallèle massif et une analyse statistique de la population enrichie. Les méthodes peuvent être utilisées avec des échantillons d'ADN provenant d'un patient, tel qu'une biopsie de tissu ou un échantillon de plasma (biopsie liquide), pour la détection de la présence de biomarqueurs tumoraux, par exemple, à des fins de diagnostic, de criblage, de sélection de thérapie et/ou de surveillance de traitement. Des kits pour mettre en œuvre les méthodes de l'invention sont également décrits.

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