

(12) International Application Status Report

Received at International Bureau: 02 November 2017 (02.11.2017)

Information valid as of: 17 December 2018 (17.12.2018)

Report generated on: 21 July 2019 (21.07.2019)

(10) Publication number:

WO2018/068014

(43) Publication date:

12 April 2018 (12.04.2018)

(26) Publication language:

English (EN)

(21) Application Number:

PCT/US2017/055653

(22) Filing Date:

06 October 2017 (06.10.2017)

(25) Filing language:

English (EN)

(31) Priority number(s):

62/405,824 (US)

(31) Priority date(s):

07 October 2016 (07.10.2016)

(31) Priority status:

Priority document received (in compliance with PCT Rule 17.1)

(51) International Patent Classification:

G06F 19/18 (2011.01); *G06F 19/22* (2011.01)

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(54) Title (EN): SYSTEM AND METHOD FOR SECONDARY ANALYSIS OF NUCLEOTIDE SEQUENCING DATA

(54) Title (FR): SYSTÈME ET PROCÉDÉ D'ANALYSE SECONDAIRE DE DONNÉES DE SÉQUENÇAGE DE NUCLÉOTIDES

(57) Abstract:

(EN): Disclosed herein are systems and methods for performing secondary analyses of nucleotide sequencing data in a time-efficient manner. Some embodiments include performing a secondary analysis iteratively while sequence reads are generated by a sequencing system. Secondary analyses can encompass both alignment of sequence reads to a reference sequence (e.g., the human reference genome sequence) and utilization of this alignment to detect differences between a sample and the reference. Secondary analysis can enable detection of genetic differences, variant detection and genotyping, identification of single nucleotide polymorphisms (SNPs), small insertions and deletion (indels) and structural changes in the DNA, such as copy number variants (CNVs) and chromosomal rearrangements.

(FR): L'invention concerne des systèmes et des procédés permettant d'effectuer des analyses secondaires de données de séquençage de nucléotides d'une manière efficace et rapide. Certains modes de réalisation consistent à effectuer une analyse secondaire de manière itérative tandis que des lectures de séquence sont générées par un système de séquençage. Des analyses secondaires peuvent englober à la fois l'alignement de lectures de séquence avec une séquence de référence (par exemple, la séquence du génome de référence humain) et l'utilisation de cet alignement pour détecter des différences entre un échantillon et la référence. L'analyse secondaire peut permettre la détection de différences génétiques, la détection de variantes et le génotypage, l'identification de polymorphismes mononucléotidiques (SNP), des petites insertions et délétions (indels) et des modifications structurales dans l'ADN, telles que des variantes du nombre de copies (CNV) et des réarrangements chromosomiques.

International search report:

Received at International Bureau: 29 January 2018 (29.01.2018) [EP]

International Report on Patentability (IPRP) Chapter II of the PCT:

Not available

(81) Designated States:

AE, AG, AL, AM, AO, AT, AU, AZ, BA, BB, BG, BH, BN, BR, BW, BY, BZ, CA, CH, CL, CN, CO, CR, CU, CZ, DE, DJ, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JO, JP, KE, KG, KH, KN, KP, KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PA, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, SA, SC, SD, SE, SG, SK, SL, SM, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, ZA, ZM, ZW

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

African Intellectual Property Organization (OAPI) : BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, KM, ML, MR, NE, SN, TD, TG

African Regional Intellectual Property Organization (ARIPO) : BW, GH, GM, KE, LR, LS, MW, MZ, NA, RW, SD, SL, ST, SZ, TZ, UG, ZM, ZW

Eurasian Patent Organization (EAPO) : AM, AZ, BY, KG, KZ, RU, TJ, TM