



- (51) **International Patent Classification:**
G06F 19/22 (2011.01) *G06F 19/28* (2011.01)
- (21) **International Application Number:**
PCT/IB2015/051278
- (22) **International Filing Date:**
19 February 2015 (19.02.2015)
- (25) **Filing Language:** English
- (26) **Publication Language:** English
- (30) **Priority Data:**
1402996.1 20 February 2014 (20.02.2014) GB
- (71) **Applicant:** VELA OPERATIONS PTE. LTD. [SG/SG];
#02-04/05/06 The Kendall, 50 Science Park Road, Singapore Science Park II, Singapore 117406 (SG).
- (72) **Inventors:** ARIYARATNE, Pramila; Blk 526 Bukit Batok Street 51, #04-114, Singapore 650526 (SG). LEE, Charlie; Blk 96 Henderson Road, #18-68, Singapore 152075 (SG).
- (81) **Designated States** (*unless otherwise indicated, for every kind of national protection available*): 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, DK, DM, DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IR, IS, JP, KE, KG, KN, KP, KR, 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.

- (84) **Designated States** (*unless otherwise indicated, for every kind of regional protection available*): ARIPO (BW, GH, GM, KE, LR, LS, MW, MZ, NA, RW, SD, SL, ST, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, RU, TJ, TM), European (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), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, KM, ML, MR, NE, SN, TD, TG).

Published:

- with international search report (Art. 21(3))
- before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments (Rule 48.2(h))

(54) **Title:** VARIANT ANALYSIS IN HIGH-THROUGHPUT SEQUENCING APPLICATIONS

(57) **Abstract:** The present invention relates to methods of determining, identifying, detecting and annotating nucleic acid sequences suspected in a sample. Particularly, the methods of the invention permit annotating nucleic acid variants present in a sample. The methods are based on data obtained in high-throughput sequencing of nucleic acids obtained from a sample. The invention relates also to computer programs capable of executing the inventive methods and apparatus suitable of running respective computer software.

