



- (51) **International Patent Classification:**
G16B 40/20 (2019.01) *G16B 20/20* (2019.01)
- (21) **International Application Number:**
PCT/US2019/048999
- (22) **International Filing Date:**
30 August 2019 (30.08.2019)
- (25) **Filing Language:** English
- (26) **Publication Language:** English
- (30) **Priority Data:**
62/726,182 31 August 2018 (31.08.2018) US
62/823,578 25 March 2019 (25.03.2019) US
62/857,048 04 June 2019 (04.06.2019) US
- (71) **Applicant:** **GUARDANT HEALTH, INC.** [US/US]; 505 Penobscot Drive, Redwood City, California 94063 (US).
- (72) **Inventors:** **ARTSIOMENKA, Aliaksandr**; 505 Penobscot Drive, Redwood City, California 94063 (US). **SIKORA, Marcin**; 505 Penobscot Drive, Redwood City, California 94063 (US). **BARBACIORU, Catalin**; 505 Penobscot Drive, Redwood City, California 94063 (US). **CHUDOVA, Darya**; 505 Penobscot Drive, Redwood City, California 94063 (US). **LEFTEROVA, Martina I.**; 505 Penobscot Drive, Redwood City, California 94063 (US).

- (74) **Agent:** **KANAKARAJ, Indhu** et al.; 505 Penobscot Drive, Redwood City, California 94063 (US).

- (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, 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.

- (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).

- (54) **Title:** MICROSATELLITE INSTABILITY DETECTION IN CELL-FREE DNA

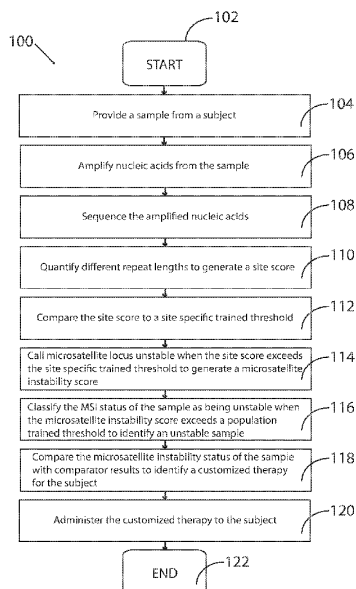


FIGURE 1

- (57) **Abstract:** Provided herein are methods for determining the microsatellite instability status of samples. In one aspect, the methods include quantifying a number of different repeat lengths present at each of a plurality of microsatellite loci from sequence information to generate a site score for each of the plurality of the microsatellite loci. The methods also include comparing the site score of a given microsatellite locus to a site specific trained threshold for the given microsatellite locus for each of the plurality of the microsatellite loci and calling the given microsatellite locus as being unstable when the site score of the given microsatellite locus exceeds the site specific trained threshold for the given microsatellite locus to generate a microsatellite instability score, which includes a number of unstable microsatellite loci from the plurality of the microsatellite loci.

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))*