



- (51) **International Patent Classification:**
G16B 20/20 (2019.01) *G16B 40/20* (2019.01)
- (21) **International Application Number:**
PCT/US2021/042605
- (22) **International Filing Date:**
21 July 2021 (21.07.2021)
- (25) **Filing Language:** English
- (26) **Publication Language:** English
- (30) **Priority Data:**
63/055,731 23 July 2020 (23.07.2020) US
- (71) **Applicant:** **ILLUMINA, INC.** [US/US]; 5200 Illumina Way, San Diego, California 92122 (US).
- (72) **Inventors:** **GAO, Hong**; 5200 Illumina Way, San Diego, California 92122 (US). **FARH, Kai-How**; 5200 Illumina Way, San Diego, California 92122 (US). **MCRAE, Jeremy, Francis**; 5200 Illumina Way, San Diego, California 92122 (US).
- (74) **Agent:** **RARIDEN, John** et al.; P.O. Box 692289, Houston, Texas 77269 (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, IT, 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, ST, SV, SY, TH, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, WS, 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).

Declarations under Rule 4.17:

- as to applicant's entitlement to apply for and be granted a patent (Rule 4.17(ii))
- as to the applicant's entitlement to claim the priority of the earlier application (Rule 4.17(iii))

- (54) **Title:** VARIANT PATHOGENICITY SCORING AND CLASSIFICATION AND USES THEREOF

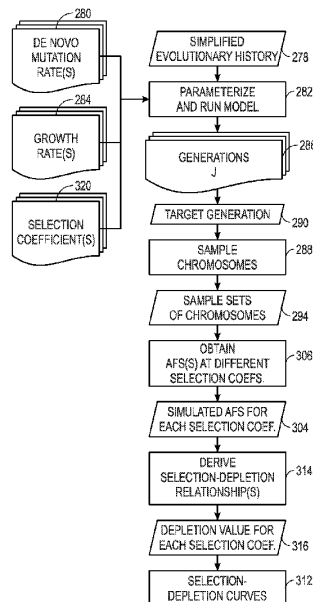


FIG. 14

- (57) **Abstract:** Derivation and use of pathogenicity scores (206) for gene variants are described herein. Applications, uses, and variations of the pathogenicity scoring process include, but are not limited to, the derivation and use of thresholds (212, 218) to characterize a variant as pathogenic or benign, the estimation of selection effects associated with a gene variant, the estimation of genetic disease prevalence using pathogenicity scores (206), and the recalibration of methods used to assess pathogenicity scores (206).

Published:

— with international search report (Art. 21(3))