



(51) International Patent Classification:

G06F 17/24 (2006.01) G06F 19/28 (2011.01)
G06F 17/30 (2006.01) G16B 20/00 (2019.01)
G06F 19/18 (2011.01) G16B 20/20 (2019.01)

(21) International Application Number:

PCT/US2021/032230

(22) International Filing Date:

13 May 2021 (13.05.2021)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

63/024,730 14 May 2020 (14.05.2020) US

(71) Applicant: **AMPEL BIOSOLUTIONS, LLC** [US/US];
250 West Main Street, Ste. 300, Charlottesville, Virginia
22902 (US).

(72) Inventors: **OWEN, Katherine A.**; 250 West Main Street,
Ste. 300, Charlottesville, Virginia 22902 (US). **BELL,
Kristy A.**; 250 West Main Street, Ste. 300, Charlottesville,
Virginia 22902 (US). **KAIN, Jessica**; 250 West Main
Street, Ste. 300, Charlottesville, Virginia 22902 (US).
GRAMMER, Amrie C.; 250 West Main Street, Ste. 300,
Charlottesville, Virginia 22902 (US). **LIPSKY, Peter E.**;
250 West Main Street, Ste. 300, Charlottesville, Virginia
22902 (US).

(74) Agent: **CHANG, Ardith**; EVERSLEDs SUTHERLAND
(US) LLP, 12255 El Camino Real, Suite 100, San Diego,
California 92130 (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,

(54) Title: METHODS AND SYSTEMS FOR MACHINE LEARNING ANALYSIS OF SINGLE NUCLEOTIDE POLYMORPHISMS IN LUPUS

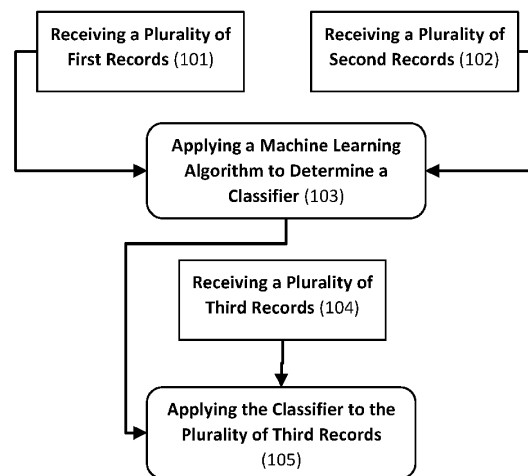


FIG. 1

(57) Abstract: The present disclosure provides systems and methods for machine learning classification and assessment of disease based on gene expression data. In an aspect, a method for determining a disease state of a subject may comprise: (a) assaying a biological sample obtained or derived from the subject to produce a data set comprising gene expression measurements of the biological sample at each of a plurality of disease-associated genomic loci; (b) computer processing the data set to determine the disease state of the subject; and (c) electronically outputting a report indicative of the disease state of the subject. In some embodiments, the plurality of disease-associated genomic loci comprises single nucleotide polymorphisms (SNPs). In some embodiments, the disease comprises a lupus condition. In some embodiments, the disease comprises cardiovascular disease (CVD).

[Continued on next page]

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))*

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))*

(88) Date of publication of the international search report:

16 December 2021 (16.12.2021)