

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property
Organization
International Bureau

(43) International Publication Date
30 November 2023 (30.11.2023)



(10) International Publication Number
WO 2023/229999 A1

(51) International Patent Classification:

C12Q 1/6806 (2018.01) C12Q 1/6869 (2018.01)
C12N 9/12 (2006.01) C40B 40/06 (2006.01)
C12Q 1/6827 (2018.01) C40B 50/06 (2006.01)

(21) International Application Number:

PCT/US2023/023113

(22) International Filing Date:

22 May 2023 (22.05.2023)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

63/345,364 24 May 2022 (24.05.2022) US

(71) Applicant: **ACCURAGEN HOLDINGS LIMITED**; P.O.
Box 472, Harbour Place, 2nd Floor, 103 South Church
Street, George Town, Grand Cayman, KY1-1106 (KY).

(72) Inventor; and

(71) Applicant (for MW only): **WENG, Li** [CN/US]; 33991
Shlylock Drive, Fremont, California 94555 (US).

(72) Inventors: **WITTKOP, Tobias**; 821 N. Delaware Street,
San Mateo, California 94401 (US). **FAHAM, Malek**; 1140
Balboa Avenue, Burlingame, California 94010 (US).

(74) Agent: **FLOYD, Jennifer A.** et al.; Wilson Sonsini, 650
Page Mill Road, Palo Alto, California 94304 (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, CV, CZ, DE, DJ, DK, DM,
DO, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT,
HN, HR, HU, ID, IL, IN, IQ, IR, IS, IT, JM, JO, JP, KE, KG,
KH, KN, KP, KR, KW, KZ, LA, LC, LK, LR, LS, LU, LY,
MA, MD, MG, MK, MN, MU, 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, CV,
GH, GM, KE, LR, LS, MW, MZ, NA, RW, SC, 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,

(54) Title: COMPOSITIONS AND METHODS FOR DETECTING RARE SEQUENCE VARIANTS

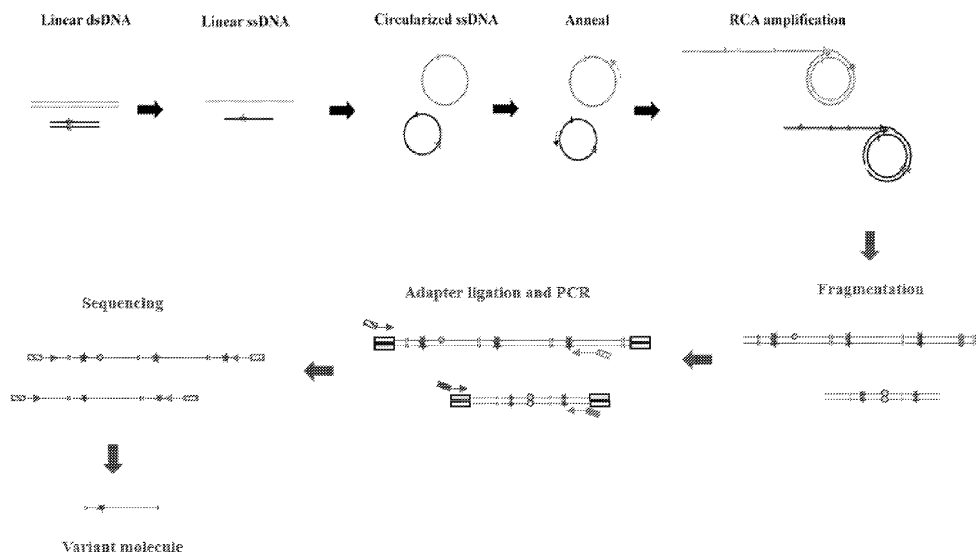


FIG. 1

(57) Abstract: Provided herein are methods and compositions for nucleic acid analysis, in particular methods for identifying sequence variants.

[Continued on next page]

LU, LV, MC, ME, 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))*