

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

(19) World Intellectual Property
Organization

International Bureau

(43) International Publication Date
07 December 2017 (07.12.2017)



(10) International Publication Number
WO 2017/206888 A1

(51) International Patent Classification:
C12Q 1/68 (2006.01)

(21) International Application Number:
PCT/CN2017/086509

(22) International Filing Date:
30 May 2017 (30.05.2017)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:
62/343,050 30 May 2016 (30.05.2016) US

(71) Applicant: THE CHINESE UNIVERSITY OF HONG KONG [CN/CN]; Office of Research and Knowledge Transfer Services, Room 301, Pi Ch'iu Building, Shatin, New Territories, Hong Kong 999077 (CN).

(72) Inventors: LO, Yuk-Ming Dennis; 4th Floor, 7 King Tak Street, Homantin, Kowloon, Hong Kong 999077 (CN). CHIU, Rossa Wai Kwun; House 31, Double Haven, 52 Ma Lok Path, Shatin, New Territories, Hong Kong 999077 (CN). CHAN, Kwan Chee; Flat C, 15th Floor, Block 5, Peak One, Mei Tin Road, Shatin, New Territories, Hong Kong 999077 (CN). SUN, Kun; Room 711, Block A, Staff Quarters, Prince of Wales Hospital, 30-32 Ngan Shing Street, Shatin, New Territories, Hong Kong 999077 (CN).

(74) Agent: INSIGHT INTELLECTUAL PROPERTY LIMITED; 19 A, Tower A, InDo Building, No.48A Zhichun Road, Haidian District, Beijing 100098 (CN).

(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, 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).

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

(54) Title: DETECTING HEMATOLOGICAL DISORDERS USING CELL-FREE DNA IN BLOOD

(57) Abstract: Techniques are provided for detecting hematological disorders using cell-free DNA in a blood sample, e.g., using plasma or serum. For example, an assay can target one or more differentially-methylated regions specific to a particular hematological cell lineage (e.g., erythroblasts). A methylation level can be quantified from the assay to determine an amount of methylated or unmethylated DNA fragments in a cell-free mixture of the blood sample. The methylation level can be compared to one or more cutoff values, e.g., that correspond to a normal range for the particular hematological cell lineage as part of determining a level of a hematological disorder.



WO 2017/206888 A1