

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

(19) World Intellectual Property
Organization
International Bureau

(43) International Publication Date
27 February 2020 (27.02.2020)



(10) International Publication Number
WO 2020/040696 A1

(51) International Patent Classification:

G01N 27/26 (2006.01) G01N 33/48 (2006.01)
C12Q 1/686 (2018.01)

(21) International Application Number:

PCT/SG2019/050410

(22) International Filing Date:

20 August 2019 (20.08.2019)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

10201807097T 21 August 2018 (21.08.2018) SG
10201809765W 02 November 2018 (02.11.2018) SG

(71) Applicants: **A. MENARINI BIOMARKERS SINGAPORE PTE LTD** [SG/SG]; 30 Pasir Panjang Road, #08-32, Mapletree Business City, Singapore 117440 (SG). **SINGAPORE HEALTH SERVICES PTE LTD** [SG/SG]; 31 Third Hospital Avenue, #03-03 Bowyer Block C, Singapore 168753 (SG).

(72) Inventors: **TAN, Wei Jian**; Blk 418 Clementi Ave 1, #20-253, Singapore 120418 (SG). **WANG, Meng**; 30 Holland Close, #09-197, Singapore 270030 (SG). **CASTAGNOLI, Paola Ricciardi**; 2 Marina Boulevard #34-05, The Sail at Marina Bay, Singapore 018987 (SG). **CHAN, Anita Sook Yee**; 4, Sixth Crescent, Singapore 276421 (SG). **LIM, Tong Seng**; Blk 211 Bukit Batok Street 21, #11-234, Singapore 650211 (SG).

(74) Agent: **DOWSING, Bruce John**; Marks & Clerk Singapore LLP, Tanjong Pagar, P O Box 636, Singapore 910816 (SG).

(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.

(54) Title: SINGLE CELLULAR DIAGNOSIS OF PRIMARY VITREORETINAL LYMPHOMA

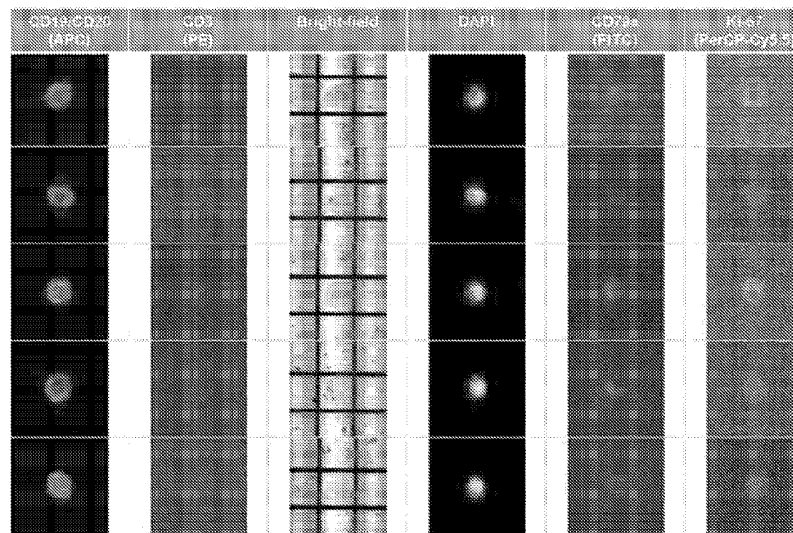


FIGURE 4

(57) Abstract: This invention relates to a methodology for single cell-based analysis of vitreous aspiration samples for diagnosis of vitreoretinal lymphoma (PVRL) such as primary vitreoretinal lymphoma (PVRL). Analysis of clonality, copy number aberration, BCL2/JH (t14;18) translocation and/or MYD88 mutations are performed on whole genomic amplified DNA derived from single B-cells to provide molecular-based diagnosis of VRL, which cannot be achieved by conventional cytology diagnosis approach.

[Continued on next page]

WO 2020/040696 A1

(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))
- with sequence listing part of description (Rule 5.2(a))