

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

(19) World Intellectual Property
Organization
International Bureau

(43) International Publication Date
08 December 2022 (08.12.2022)



(10) International Publication Number
WO 2022/255952 A3

(51) International Patent Classification:
C12Q 1/6858 (2018.01) *C12Q 1/6883* (2018.01)

(21) International Application Number:
PCT/SG2022/050380

(22) International Filing Date:
02 June 2022 (02.06.2022)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:
10202105851T 02 June 2021 (02.06.2021) SG

(71) Applicants: NATIONAL UNIVERSITY OF SINGAPORE [SG/SG]; 21 Lower Kent Ridge Road, Singapore 119077 (SG). NATIONAL UNIVERSITY HOSPITAL (SINGAPORE) PTE LTD [SG/SG]; 1E Kent Ridge Road, #13-00, Singapore 119228 (SG).

(72) Inventors: **CHONG, Siong-Chuan Samuel**; c/o National University of Singapore, Yong Loo Lin School of Medicine / Department of Paediatrics, 21 Lower Kent Ridge Road, Singapore 119077 (SG). **LEE, Guat Lay, Caroline**; c/o National University of Singapore, Yong Loo Lin School of Medicine / Department of Biochemistry, 21 Lower Kent Ridge Road, Singapore 119077 (SG). **WANG, Sijia Furene**; c/o National University of Singapore, Yong Loo Lin School of Medicine / Department of Paediatrics, 21 Lower Kent Ridge Road, Singapore 119077 (SG). **LIU, Timing**; c/o National University of Singapore, Yong Loo Lin School of Medicine / Department of Paediatrics, 21 Lower Kent Ridge Road, Singapore 119077 (SG). **CHEAH, Siew Hong**; c/o National University Health System, 1E Kent Ridge Road, Level 12, NUHS Tower Block, Singapore 119228 (SG). **LIAN, Mulias Ya Hui**; c/o National University Health System, 1E Kent Ridge Road, Level 12, NUHS Tower Block, Singapore 119228 (SG).

(54) Title: METHOD OF DETECTING A REPEAT EXPANSION SEQUENCE

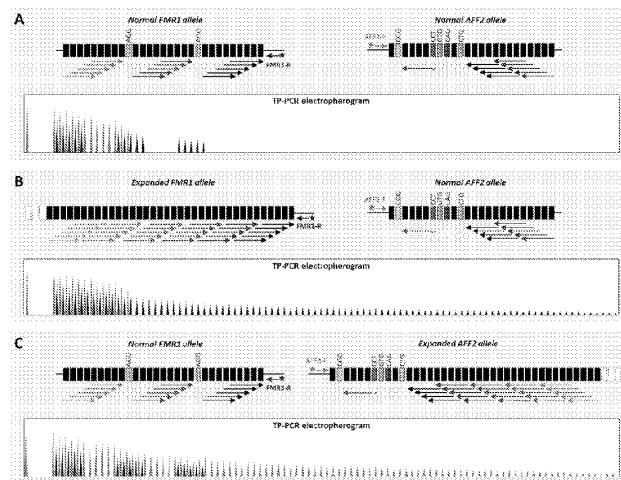


Figure 1

(57) Abstract: The present disclosure relates, inter alia, a method of detecting the presence or absence of a repeat expansion sequence in two or more genes in a nucleic acid sample obtained from a subject, the method comprising i) contacting the nucleic acid sample for each of the two or more genes with a) a gene-specific primer that binds to a distinct target sequence of a respective gene wherein the gene comprises a nucleotide repeat sequence and wherein the distinct target sequence is upstream or downstream of the nucleotide repeat sequence, and b) a common primer that binds to a common target sequence that is shared by the two or more genes, wherein the common target sequence is located within the nucleotide repeat sequence and on the opposite strand that is bound by the gene-specific primer; and ii) analyzing the amplification products.

[Continued on next page]

(74) **Agent: DAVIES COLLISON CAVE ASIA PTE. LTD.;**
10 Collyer Quay #07-01, Ocean Financial Centre, Singapore 049315 (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, IQ, IR, IS, IT, JM, 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).

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

(88) **Date of publication of the international search report:**
12 January 2023 (12.01.2023)