

(19) World Intellectual Property Organization
International Bureau



(43) International Publication Date
12 February 2004 (12.02.2004)

PCT

(10) International Publication Number
WO 2004/012668 A3

(51) International Patent Classification:

C07K 14/00 (2006.01) **C12N 15/11** (2006.01)
C07K 16/00 (2006.01) **C12N 15/63** (2006.01)
C12N 5/10 (2006.01) **G01N 33/53** (2006.01)

(21) International Application Number:

PCT/US2003/024190

(22) International Filing Date: 1 August 2003 (01.08.2003)

(25) Filing Language: English

(26) Publication Language: English

(30) Priority Data:

60/401,018 2 August 2002 (02.08.2002) US

(81) Designated States (*national*): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States (*regional*): ARIPO patent (GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE, SI, SK, TR), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

(71) Applicant: **WISCONSIN ALUMNI RESEARCH FOUNDATION** [US/US]; 614 Walnut Street, P.O. Box 7365, Madison, WI 53707-7365 (US).

(72) Inventors: **MAKIELSKI, Jonathan, C.**; 2738 Richardson Street, Fitchburg, WI 53711 (US). **YE, Bin**; 939 Schroll Drive, Forsyth, IL 62535 (US). **ACKERMAN, Michael, J.**; 716 4th Street SW, Rochester, MN 55902 (US).

(74) Agent: **BERSON, Benneth, J.**; Quarles & Brady LLP, P.O. Box 2113, Madison, WI 53701-2113 (US).

Published:

- with international search report
- before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments

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

7 December 2006

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

(54) Title: SODIUM CHANNEL ALPHA SUBUNIT VARIANTS

(57) Abstract: The present invention discloses four groups of SCN5A variants that represent the most common SCN5A variants in humans. A specific mutation in one of the variants has been shown to display a different phenotype in relation to a human heart disease than other variants and known human sodium channel α subunits with corresponding mutations. The present invention provides new tools to study mutations and to design or identify new diagnostic and treatment strategies or agents for sodium channel related diseases or conditions.



WO 2004/012668 A3

INTERNATIONAL SEARCH REPORT

I

PCT/US03/24190

A. CLASSIFICATION OF SUBJECT MATTER

IPC: C07K 14/00(2006.01),16/00(2006.01);C12N 5/10(2006.01),15/11(2006.01),15/63(2006.01);G01N 33/53(2006.01)

USPC: 435/6,7.2,320.1,325;530/350,387.1;536/23.5

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

U.S. : 435/6, 7.2, 320.1, 325; 530/350, 387.1; 536/23.5

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practicable, search terms used)
Please See Continuation Sheet

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	US 5,599,673 (KEATING et al.) 04 February 1997 (04.02.1997) see entire document.	1-50

☐ Further documents are listed in the continuation of Box C.

☐ See patent family annex.

* Special categories of cited documents:

"A" document defining the general state of the art which is not considered to be of particular relevance

"E" earlier application or patent published on or after the international filing date

"L" document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)

"O" document referring to an oral disclosure, use, exhibition or other means

"P" document published prior to the international filing date but later than the priority date claimed

"T" later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention

"X" document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone

"Y" document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art

"&" document member of the same patent family

Date of the actual completion of the international search

19 September 2006 (19.09.2006)

Date of mailing of the international search report

05 OCT 2006

Name and mailing address of the ISA/US

Mail Stop PCT, Attn: ISA/US
Commissioner of Patents
P.O. Box 1450
Alexandria, Virginia 22313-1450

Facsimile No. (571) 273-3201

Authorized officer

Michael Pak

Telephone No. 571-272-1600

INTERNATIONAL SEARCH REPORT

PCT/US03/24180

Continuation of B. FIELDS SEARCHED Item 3:
BRS, GENESEQ, PIR, UNIPROT

search terms: sodium channel, SCN5A, variant