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- (72) Inventors; and
- (71) Applicants : EHLI, Erik [US/US]; 7404 W. 53rd Street, Sioux Falls, SD 57106 (US). DAVIES, Gareth [GB/US]; 6215 N. Lakeside Drive, Madison, SD 57042 (US). ABDELLAOUI, Abdel [NL/NL]; Vu University, Van Der Boechorststraat 1, NL-1081 BT Amsterdam (NL). HUDZIAK, James [US/US]; PO Box 547, Charlotte, VT 05545 (US).
- (74) Agent: NAKAMURA, Dean; MDIP LLC, Post Office Box 2630, Montgomery Village, MD 20886 (US).
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[Continued on next page]

(54) Title: METHOD OF IDENTIFYING *DE NOVO* COPY NUMBER VARIANTS (CNV) USING MZ TWINS DISCORDANT FOR ATTENTION PROBLEMS/DISORDERS

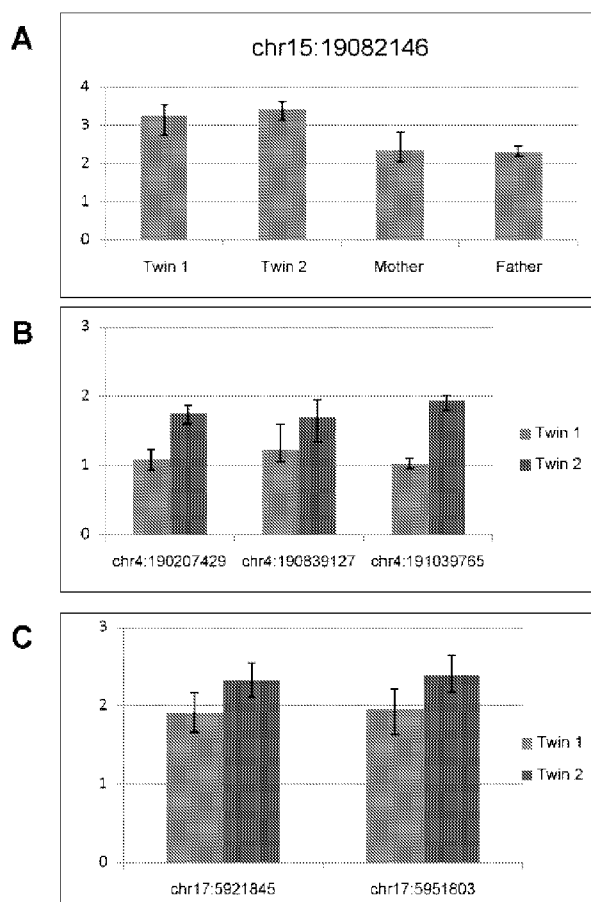


FIGURE 4

(57) Abstract: The present disclosure relates to methods of identifying copy number variation (CNV) events and sizes that are associated with attention problems (AP) and identifying one or more CNVs which correlate with such disorders.





DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, GT, HN, HR, HU, ID, IL, IN, IS, JP, KE, KG, KM, KN, KP, KR, KZ, LA, LC, LK, LR, LS, LT, LU, LY, MA, MD, ME, MG, MK, MN, MW, MX, MY, MZ, NA, NG, NI, NO, NZ, OM, PE, PG, PH, PL, PT, QA, RO, RS, RU, RW, 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.

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**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))

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

4 April 2013

**A. CLASSIFICATION OF SUBJECT MATTER***C12Q 1/68(2006.01)i*

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)

C12Q 1/68; C12N 5/06; C40B 40/06; C07H 21/00; G06N 5/02

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

Korean utility models and applications for utility models

Japanese utility models and applications for utility models

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

eKOMPASS(KIPO internal) &amp; Keywords: copy number variants, CNV, attention disorder, AD, monozygotic twins, MZ, contrast quality control, median absolute pairwise difference, HERC2P3, HSP90AA4P, FRG1, TUBB4Q, FRG2, DUX4, WSCD1, etc.

**C. DOCUMENTS CONSIDERED TO BE RELEVANT**

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	US 2011-0111419 A1 (HREINN STEFANSSON et al.) 12 May 2011 See claims 1 and 30; paragraphs [0018], [0085], [0276], and [0284].	11, 13-20
A	US 2010-0248235 A1 (STEPHEN W. SCHERER et al.) 30 September 2010 See claim 30.	11, 13-20
A	WO 2009-105718 A1 (THE CHILDREN'S HOSPITAL OF PHILADELPHIA) 27 August 2009 See claims 1 and 3; table 1.	11, 13-20
A	US 2009-0307180 A1 (BRANDON COLBY et al.) 10 December 2009 See claim 1.	11, 13-20
A	US 2010-0143921 A1 (WOLFGANG SADEE et al.) 10 June 2010 See claim 1; paragraph [0305].	11, 13-20
PX	ERIK A. EHLI et al., 'De novo and inherited CNVs in MZ twin pairs selected for discordance and concordance on Attention Problems', European Journal of Human Genetics, 11 April 2012, Vol. 20, No. 10, pages 1037-1043, ISSN 1018-4813. See whole document.	11, 13-20

 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 but 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 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

"&amp;" document member of the same patent family

Date of the actual completion of the international search

30 JANUARY 2013 (30.01.2013)

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**31 JANUARY 2013 (31.01.2013)**

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Korean Intellectual Property Office  
189 Cheongsa-ro, Seo-gu, Daejeon Metropolitan  
City, 302-701, Republic of Korea

Facsimile No. 82-42-472-7140

Authorized officer

Heo, Joo-Hyung

Telephone No. 82-42-481-8150



**Box No. II Observations where certain claims were found unsearchable (Continuation of item 2 of first sheet)**

This international search report has not been established in respect of certain claims under Article 17(2)(a) for the following reasons:

1.  Claims Nos.: 1-7  
because they relate to subject matter not required to be searched by this Authority, namely:  
Claims 1-7 pertain to methods for treatment of the human body by surgery, as well as diagnostic methods, and thus relate to a subject matter which this International Searching Authority is not required, under Article 17(2)(a)(i) of the PCT and Rule 39.1(iv) of the Regulations under the PCT, to search.
2.  Claims Nos.: 8-10, 12  
because they relate to parts of the international application that do not comply with the prescribed requirements to such an extent that no meaningful international search can be carried out, specifically:  
The technical features in the claims 8-10 and 12 relate to a method of identifying copy number variants (CNV) rather than clearly defining the CNV in terms of its technical features. Therefore, the intended limitations are not clear from said claims, contrary to the requirements of PCT Article 6.
3.  Claims Nos.:  
because they are dependent claims and are not drafted in accordance with the second and third sentences of Rule 6.4(a).

**Box No. III Observations where unity of invention is lacking (Continuation of item 3 of first sheet)**

This International Searching Authority found multiple inventions in this international application, as follows:

1.  As all required additional search fees were timely paid by the applicant, this international search report covers all searchable claims.
2.  As all searchable claims could be searched without effort justifying an additional fee, this Authority did not invite payment of any additional fee.
3.  As only some of the required additional search fees were timely paid by the applicant, this international search report covers only those claims for which fees were paid, specifically claims Nos.:
  
4.  No required additional search fees were timely paid by the applicant. Consequently, this international search report is restricted to the invention first mentioned in the claims; it is covered by claims Nos.:

**Remark on Protest**

- The additional search fees were accompanied by the applicant's protest and, where applicable, the payment of a protest fee.
- The additional search fees were accompanied by the applicant's protest but the applicable protest fee was not paid within the time limit specified in the invitation.
- No protest accompanied the payment of additional search fees.

**INTERNATIONAL SEARCH REPORT**

Information on patent family members

International application No.

**PCT/US2012/040795**

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