



US011072830B2

(12) **United States Patent**
Hinds et al.

(10) **Patent No.:** US 11,072,830 B2
(b4) **Date of Patent:** Jul. 27, 2021

(54) **METHODS FOR BREAST CANCER RISK ASSESSMENT**(71) Applicant: **Genetic Technologies Limited**, Fitzroy (AU)(72) Inventors: **David A. Hinds**, Sunnyvale, CA (US); **Bryan Walser**, Menlo Park, CA (US)(73) Assignee: **GENETIC TECHNOLOGIES LIMITED**, Fitzroy (AU)

(*) Notice: Subject to any disclaimer, the term of this patent is extended or adjusted under 35 U.S.C. 154(b) by 0 days.

(21) Appl. No.: **15/814,255**(22) Filed: **Nov. 15, 2017**(65) **Prior Publication Data**

US 2018/0080089 A1 Mar. 22, 2018

Related U.S. Application Data

(63) Continuation of application No. 12/920,815, filed as application No. PCT/AU2010/000675 on Jun. 1, 2010, now abandoned.

(60) Provisional application No. 61/258,420, filed on Nov. 5, 2009, provisional application No. 61/182,809, filed on Jun. 1, 2009.

(51) **Int. Cl.**
C12Q 1/6886 (2018.01)(52) **U.S. Cl.**
CPC **C12Q 1/6886** (2013.01); **C12Q 2600/106** (2013.01); **C12Q 2600/112** (2013.01); **C12Q 2600/156** (2013.01); **C12Q 2600/172** (2013.01)(58) **Field of Classification Search**

None

See application file for complete search history.

(56) **References Cited**

U.S. PATENT DOCUMENTS

6,812,339	B1	11/2004	Venter
7,127,355	B2	10/2006	Cox et al.
7,529,685	B2	5/2009	Davies et al.
9,051,617	B2	6/2015	Cox
2002/0123095	A1*	9/2002	Kalush
			C07K 14/70567 435/69.1
2003/0092019	A1	5/2003	Meyer
2003/009964	A1	5/2003	Patil
2003/0108910	A1	6/2003	Toland
2003/0157488	A1	8/2003	Cox
2004/0002071	A1	1/2004	Ralph
2004/0023237	A1	2/2004	Patil
2004/0191783	A1*	9/2004	Leclercq
			C12Q 1/6837 506/16
2004/0210400	A1	10/2004	Konvicka
2004/0229224	A1	11/2004	Frazer
2004/0241657	A1	12/2004	Patil
2005/0003410	A1	1/2005	Frazer
2005/0019787	A1	1/2005	Berno
2005/0084849	A1	4/2005	Moskowitz

2005/0196770	A1	9/2005	Cox
2007/0037198	A1	2/2007	Cox et al.
2007/0166738	A1	7/2007	Cox et al.
2008/0131887	A1	6/2008	Stephan et al.
2009/0087854	A1	4/2009	Cox et al.
2009/0208962	A1	8/2009	Cox et al.
2009/0239226	A1	9/2009	Cox et al.
2009/0239763	A1	9/2009	Cox et al.
2011/0015081	A1*	1/2011	Stacey
			C12Q 1/6886 506/7
2011/0015092	A1	1/2011	Cox et al.
2011/0200588	A1	8/2011	Cox et al.
2011/0294681	A1	12/2011	Hinds et al.
2016/0222469	A1	8/2016	Allman
2017/0275707	A1	9/2017	Cox
2018/0305764	A1	10/2018	Allman

FOREIGN PATENT DOCUMENTS

WO	WO 95/011995	5/1995
WO	WO 98/020167	5/1998
WO	WO 2003/025141	3/2003
WO	WO 2005/014846	2/2005
WO	WO 2005/024067	3/2005
WO	WO 00/33161	6/2005
WO	WO 2005/086770	9/2005
WO	2007/064776	6/2007
WO	WO 2007/150044	12/2007
WO	2008/117314	10/2008
WO	2008/146309	12/2008
WO	WO 2009/009752	1/2009
WO	WO 2009/097270	8/2009
WO	WO 2010/017520	2/2010
WO	WO 2010/030929	3/2010
WO	WO 2010/139006	9/2010
WO	WO 2013/151413	10/2013

OTHER PUBLICATIONS

Lee et al. (Cancer Epidemiol Biomarkers Prev Jan. 2008 vol. 17 p. 258) (Year: 2008).
Gail et al. (JNCI 2008 VO. 100 p. 1037) (Year: 2008).
Millikan et al. (Cancer Epidemiology, Biomarkers and Prevention 2005 vol. 14 p. 2326) (Year: 2005).
Mcinerney et al. (Breast Cancer Res Treat Nov. 13, 2008 vol. 117 p. 151) (Year: 2008).
Jun. 11, 2012 Office Action dated in connection with U.S. Appl. No. 12/920,815.
Jul. 11, 2012 Response filed in connection with U.S. Appl. No. 12/920,815.

(Continued)

Primary Examiner — Katherine D Salmon

(74) Attorney, Agent, or Firm — Gary J. Gershik

(57) **ABSTRACT**

The present invention relates to methods and systems for assessing the overall risk of a human female subject for developing a breast cancer phenotype. In particular, the present invention relates to combining clinical risk assessment and genetic risk assessment to improve risk analysis.

20 Claims, 2 Drawing Sheets

Specification includes a Sequence Listing.

(56)

References Cited**OTHER PUBLICATIONS**

- Sep. 21, 2012 Office Action issued in connection with U.S. Appl. No. 12/920,815.
- Feb. 21, 2013 Response filed in connection with U.S. Appl. No. 12/920,815.
- Jun. 24, 2013 Office Action issued in connection with U.S. Appl. No. 12/920,815.
- Dec. 23, 2013 Response filed in connection with U.S. Appl. No. 12/920,815.
- Apr. 20, 2015 Office Action issued in connection with U.S. Appl. No. 12/920,815.
- Oct. 20, 2015 Response filed in connection with U.S. Appl. No. 12/920,815.
- Jun. 14, 2016 Office Action issued in connection with U.S. Appl. No. 12/920,815.
- Dec. 14, 2016 Response filed in connection with U.S. Appl. No. 12/920,815.
- Jun. 19, 2017 Office Action issued in connection with U.S. Appl. No. 12/920,815.
- Extended European Search Report, dated Oct. 18, 2012 in connection with European Patent Application No. 10782820.4.
- European Search Opinion, dated Oct. 18, 2012 in connection with European Patent Application No. 10782820.4.
- Patent Examination Report No. 1, dated Oct. 9, 2012 in connection with Australian Patent Application No. 2010256343.
- Anonymous, "Perlegen Builds Commercial Group to Support Upcoming MammaPLUS™ Launch" Evaluatepharma Press Release, Apr. 28, 2009.
- Anonymous, "Perlegen Introduces BRAVAGen™ Breast Cancer Risk Stratification Test" Medical News Today, Sep. 24, 2009.
- Bondy et al. (1994) "Validation of a Breast Cancer Risk Assessment Model in Women With a Positive Family History" Journal of the National Cancer Institute, vol. 86, No. 8, pp. 620-625.
- Costantino et al. (1999) "Validation Studies for Models Projecting the Risk of Invasive and Total Breast Cancer Incidence" Journal of the National Cancer Institute, vol. 91, No. 18, pp. 1541-1548.
- Jope et al. (2007) "The OncoVue (R) model for predicting breast cancer risk" Breast Cancer Research and Treatment vol. 106 No. Suppl. 1, p. S179.
- Matsuno et al. (2011) "Projecting Individualized Absolute Invasive Breast Cancer Risk in Asian and Pacific Islander American Women" J Natl Cancer Inst 2011;13:951-961.
- Schonfeld et al. (2010) "Effect of Changing Breast Cancer Incidence Rates on the Calibration of the Gail Model" J Clin Oncol 28, pp. 1-8.
- Spiegelman et al. (1994) "Validation of the Gail et al. Model for Predicting Individual Breast Cancer Risk" Journal of the National Cancer Institute 68(8):600-607.
- Zheng et al. (2010) "Genetic and Clinical Predictors for Breast Cancer Risk Assessment and Stratification Among Chinese Women" Journal of the National Cancer Institute 102(13):972-981.
- Liu et al. (2006) "The role of Self-Defined Race/Ethnicity in Population Structure Control" Annals of Human Genetics 70:496-505.
- Merriam-Webster Dictionary definition for "Caucasian," available from www.merriam-webster.com/medical/Caucasian, accessed Nov. 2, 2012.
- Nov. 25, 2013 Office Action, issued in connection with U.S. U.S. Appl. No. 12/370,972.
- Breast Cancer. American Cancer Society. 2013, pp. 1-134. Downloaded from Cancer.org on Oct. 17, 2013.
- Rodi et al. (2003) "A Strategy for the Rapid Discovery of Disease Markers Using the MassARRAYTM System." BioTechniques, 32:S62-S69.
- Db SNP ss No. 23605537 (www.ncbi.nlm.nih.gov/projects/SNP/snp_ss.cgi?subspn_id=23605537) Aug. 20, 2004.
- Db SNP ss No. 23575498 (www.ncbi.nlm.nih.gov/SNP/snp_ss.cgi?subspn_id=23575498) Aug. 20, 2004.
- Db SNP ss No. 24561224 (www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs3817198) Aug. 20, 2004.
- Armstrong et al. (2000) "Assessing the Risk of Breast Cancer. The New England Journal of Medicine," 342(8):564-571.
- Jul. 9, 2013 Response filed in connection with U.S. Appl. No. 12/890,272.
- Jul. 10, 2013 Interview Summary issued in connection with U.S. Appl. No. 12/890,272.
- Advani and Morena-Aspitia (2014) Breast Cancer: Targets & Therapy; 6: 59-71.
- American Cancer Society: (2013) Breast Cancer Facts & Figures 2013-2014. Atlanta (GA), American Cancer Society Inc, 12.
- Brentall et al. (2014) British Journal of Cancer 110: 827-828.
- Chen et al. (2004) Stat Appl Genet Mol Biol. 3: Article 21.
- Claus et al. (1991) Am J Hum Genet. 48: 232-242.
- Claus et al. (1993) Breast Cancer Res Treat. 28: 115-120.
- Cuzick et al. (2014) Journal of Clinical Oncology 43:Abstract No. 1519.
- Darabi et al. (2012) Breast Cancer Research 14: R25.
- De la Cruz (2014) Prim Care Clin Office Pract; 41: 283-306.
- Dite et al. (2013) Breast Cancer Res Treat. 139: 887-896.
- Ellis et al. (2006) Genetic Epidemiology 30: 48-61.
- (1997a) FASEB Journal 11:A879.
- Fodor (1997b) Science 277: 393-395.
- Garcia (2004) Pediatrics 113:1394.
- Ho et al. (2013) The Malaysian Journal of Pathology 1: 33-43.
- Mackarem et al. (2001) The Breast Journal 2001 vol. 7 p. 34.
- Mahoney et al. (2008) Cancer J Clin; 58: 347-371.
- Mazzola et al. (2014) Cancer Epidemiol Biomarkers Prev. 23: 1689-1695.
- McCarthy et al. (2013) Breast Cancer Research & Treatment 138: 889-898.
- Parmigiani et al. (1998) Am J Hum Genet. 62: 145-158.
- Parmigiani et al. (2007) Ann Intern Med. 1479: 441-450.
- Raskin et al. (2008) Cancer Epidemiol Biomarkers Prev Vo. 17 p. 1060.
- Ruiz-Narvaez (2010) Breast Cancer Res Treat vol. 123 p. 525.
- Saslow et al. (2007) CA Cancer J Clin. 57: 75-89.
- Schwartz (2001) N. Engl. J. Med. 344(18):1392-1393.
- Sorlie et al. (2001) Proc. Natl. Acad. Sci., 98: 10869-10874.
- Visvanathan et al. (2009) Journal of Clinical Oncology. 27: 3235-3258.
- WIRES: Questions and Answers. "Average risk of breast cancer and general information".
- International Preliminary Report on Patentability, dated Dec. 6, 2011, in connection with PCT International Application No. PCT/AU2010/000675.
- International Search Report, dated Aug. 13, 2010 in connection with PCT International Application No. PCT/AU2010/000675.
- "Perlegen Builds Commercial Group to Support Upcoming MammaPLUS™ Launch", <URL <http://www.evaluatepharma.com/Universal/View.aspx?type=Story&id=184247>> published on Apr. 28, 2009.
- "Perlegen Introduces BREVAGen Breast Cancer Risk Stratification test" <<http://www.medicalnewstoday.com/releases/165123.php>> retrieved by the International Search Authority on Jun. 23, 2010 in connection with PCT International Application No. PCT/AU2010/000675.
- Easton et al., "Genome-wide association study identifies novel breast cancer susceptibility loci" Nature (2007) 447(7148):1087-1093.
- Ahmed et al., "Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2" Nature Genetics (2009) 41(5):585-590.
- Antoniou et al. "A comprehensive model for familial breast cancer incorporating BRCA1, BRCA2 and other genes" British Journal of Cancer (2002) 86, 76-83.
- Antoniou et al., "The BOADICEA model of genetic susceptibility to breast and ovarian cancer" British Journal of Cancer (2004) 91, 1580-1590.
- Antoniou et al., "Common variants in LSP1, 2q25 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers" Human Genetics (2009) 18(22):4442-4456.
- Barringer et al., "Blunt-end and single-strand ligations by *Escherichia coli* ligase: influence on and in vitro amplification scheme" Gene (1990) 89, 117-122.