

Title (en)
METHODS, COMPOSITIONS AND COMPUTER SOFTWARE PRODUCTS FOR INTERROGATING SEQUENCE VARIATIONS IN FUNCTIONAL GENOMIC REGIONS

Title (de)
VERFAHREN, ZUSAMMENSETZUNGEN UND COMPUTERPROGRAMME ZUR ABFRAGE VON SEQUENZVARIATIONEN IN FUNKTIONELLEN GENOMREGIONEN

Title (fr)
PROCEDES, COMPOSITIONS ET LOGICIELS INFORMATIQUES POUR INTERROGER DES VARIATIONS DE SEQUENCES DANS LES REGIONS GENOMIQUES FONCTIONNELLES

Publication
EP 1563090 A4 20070711 (EN)

Application
EP 03768853 A 20031112

Priority

- US 0335819 W 20031112
- US 42588002 P 20021112
- US 42587902 P 20021112

Abstract (en)
[origin: WO2004044700A2] In one aspect of the invention, methods, compositions and computer software products are provided to detect genetic variations in functional regions of a genome. In some embodiments, oligonucleotide tiling probe arrays are used to detect transcribed regions, transcription factor binding regions, etc. Genetic variations in these regions can be determined with, e.g., a whole genome sampling assay.

IPC 1-7
C12Q 1/68; C12P 19/34; C07H 21/04

IPC 8 full level
C12Q 1/68 (2006.01); **G16B 30/00** (2019.01)

CPC (source: EP US)
C12Q 1/6876 (2013.01 - EP); **G16B 30/00** (2019.01 - EP US)

Citation (search report)

- [DPX] US 2003186280 A1 20031002 - KENNEDY GIULIA C [US]
- [X] FAN J-B ET AL: "Parallel genotyping of human SNPs using generic high-density oligonucleotide tag arrays", GENOME RESEARCH, COLD SPRING HARBOR LABORATORY PRESS, WOODBURY, NY, US, vol. 10, no. 6, 2000, pages 853 - 860, XP002236784, ISSN: 1088-9051
- [X] WANG D G ET AL: "Large-scale identification, mapping, and genotyping of single-nucleotide polymorphisms in the human genome", SCIENCE, AMERICAN ASSOCIATION FOR THE ADVANCEMENT OF SCIENCE, US, vol. 280, 1998, pages 1077 - 1082, XP002089398, ISSN: 0036-8075
- [X] SCHUBERT E L ET AL: "Single nucleotide polymorphism array analysis of flow-sorted epithelial cells from frozen versus fixed tissues for whole genome analysis of allelic loss in breast cancer", AMERICAN JOURNAL OF PATHOLOGY, vol. 160, no. 1, January 2002 (2002-01-01), pages 73 - 79, XP008078927
- [X] CUTLER DAVID J ET AL: "High-throughput variation detection and genotyping using microarrays", GENOME RESEARCH, COLD SPRING HARBOR LABORATORY PRESS, WOODBURY, NY, US, vol. 11, no. 11, November 2001 (2001-11-01), pages 1913 - 1925, XP002311211, ISSN: 1088-9051
- [X] LINDBLAD-TOH K ET AL: "LOSS-OF-HETEROZYGOSITY ANALYSIS OF SMALL-CELL LUNG CARCINOMAS USING SINGLE-NUCLEOTIDE POLYMORPHISM ARRAYS", NATURE BIOTECHNOLOGY, NATURE PUBLISHING GROUP, NEW YORK, NY, US, vol. 18, no. 9, September 2000 (2000-09-01), pages 1001 - 1005, XP001063318, ISSN: 1087-0156
- [X] MCDONALD O G KRYNETSKI E Y EVANS W E: "Molecular haplotyping of genomic DNA for multiple single nucleotide polymorphisms located kilobases apart using long range polymerase chain reaction and intramolecular ligation", PHARMACOGENETICS, CHAPMAN & HALL, LONDON, GB, vol. 12, no. 2, February 2002 (2002-02-01), pages 93 - 99, XP002956448, ISSN: 0960-314X
- [X] WANG M ET AL: "Construction and characterization of a human chromosome 2-specific BAC library", GENOMICS, vol. 24, 1994, pages 527 - 534, XP002434029
- [X] LIU N-P ET AL: "Physical and genetic mapping of the macular corneal dystrophy locus on chromosome 16q and exclusion of TAT and LCAT as candidate genes", MOLECULAR VISION, vol. 6, 2000, pages 95 - 100, XP002434030
- [X] XIANG F I ET AL: "A Huntington disease-like neurodegenerative disorder maps to chromosome 20p", AMERICAN JOURNAL OF HUMAN GENETICS, vol. 63, 1998, pages 1431 - 1438, XP008078926
- [DPX] KENNEDY G C ET AL: "Large-scale genotyping of complex DNA", NATURE BIOTECHNOLOGY, vol. 21, no. 10, October 2003 (2003-10-01), pages 1233 - 1237, XP002434032
- See references of WO 2004044700A2

Designated contracting state (EPC)
AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HU IE IT LI LU MC NL PT RO SE SI SK TR

DOCDB simple family (publication)
WO 2004044700 A2 20040527; WO 2004044700 A3 20050506; AU 2003291453 A1 20040603; AU 2003291453 A8 20040603; CN 1711358 A 20051221; EP 1563090 A2 20050817; EP 1563090 A4 20070711

DOCDB simple family (application)
US 0335819 W 20031112; AU 2003291453 A 20031112; CN 200380103082 A 20031112; EP 03768853 A 20031112