

Title (en)

METHODS AND COMPOSITIONS FOR EVALUATING GENETIC MARKERS

Title (de)

VERFAHREN UND ZUSAMMENSETZUNG ZUR BEWERTUNG GENETISCHER MARKER

Title (fr)

PROCÉDÉS ET COMPOSITIONS POUR L'ÉVALUATION DE MARQUEURS GÉNÉTIQUES

Publication

**EP 2971114 A4 20161123 (EN)**

Application

**EP 14762322 A 20140314**

Priority

- US 201361789164 P 20130315
- US 201313934093 A 20130702
- US 2014028212 W 20140314

Abstract (en)

[origin: WO2014143994A2] Aspects of the invention relates to methods and compositions that are useful to reduce bias and increase the reproducibility of multiplex analysis of genetic loci. In some configurations, predetermined preparative steps and/or nucleic acid sequence analysis techniques are used in multiplex analyses for a plurality of genetic loci in a plurality of samples.

IPC 8 full level

**C12Q 1/68** (2006.01); **C12Q 1/02** (2006.01); **G01N 33/50** (2006.01)

CPC (source: EP)

**C12Q 1/6827** (2013.01); **C12Q 1/6844** (2013.01); **C12Q 1/6883** (2013.01); **C12Q 2600/156** (2013.01)

Citation (search report)

- [E] WO 2014074246 A1 20140515 - GOOD START GENETICS INC [US]
- [Y] US 2012165202 A1 20120628 - PORRECA GREGORY [US], et al
- [Y] ISOSOMMPI JUHA ET AL: "Disease-causing mutations in the CLRN1 gene alter normal CLRN1 protein trafficking to the plasma membrane.", MOLECULAR VISION 2009, vol. 15, 2009, pages 1806 - 1818, XP055310082, ISSN: 1090-0535
- [IP] MARK A. UMBARGER ET AL: "Next-generation carrier screening", GENETICS IN MEDICINE, vol. 16, no. 2, 13 June 2013 (2013-06-13), pages 132 - 140, XP055186040, ISSN: 1098-3600, DOI: 10.1038/gim.2013.83
- [IP] STEPHANIE HALLAM ET AL: "Validation for Clinical Use of, and Initial Clinical Experience with, a Novel Approach to Population-Based Carrier Screening using High-Throughput, Next-Generation DNA Sequencing", JOURNAL OF MOLECULAR DIAGNOSTICS, THE, vol. 16, no. 2, 27 December 2013 (2013-12-27), US, pages 180 - 189, XP055309745, ISSN: 1525-1578, DOI: 10.1016/j.jmoldx.2013.10.006
- See references of WO 2014143994A2

Designated contracting state (EPC)

AL AT BE BG CH CY CZ DE DK EE ES FI FR GB GR HR HU IE IS IT LI LT LU LV MC MK MT NL NO PL PT RO RS SE SI SK SM TR

DOCDB simple family (publication)

**WO 2014143994 A2 20140918; WO 2014143994 A3 20150219;** CA 2907177 A1 20140918; EP 2971114 A2 20160120; EP 2971114 A4 20161123

DOCDB simple family (application)

**US 2014028212 W 20140314;** CA 2907177 A 20140314; EP 14762322 A 20140314