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(54) **METHOD AND SYSTEM FOR DETERMINING WHETHER COPY NUMBER VARIATION EXISTS IN SAMPLE GENOME, AND COMPUTER READABLE MEDIUM**

VERFAHREN UND SYSTEM ZUR BESTIMMUNG DER EXISTENZ EINER KOPIEZAHLVARIANTE IN EINER GENOMPROBE UND COMPUTERLESBARES MEDIUM

PROCÉDÉ ET SYSTÈME POUR DÉTERMINER S'IL EXISTE UNE VARIABILITÉ DU NOMBRE DE COPIES DANS UN GÉNOME ÉCHANTILLON, ET SUPPORT LISIBLE PAR ORDINATEUR

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(56) References cited:  
**EP-A1- 2 772 549** **WO-A1-2011/091046**  
**WO-A2-2007/129000** **WO-A2-2007/131135**  
**WO-A2-2010/033578** **WO-A2-2012/006291**  
**US-A1- 2003 082 606**

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- ALEX S NORD ET AL: "Accurate and exact CNV identification from targeted high-throughput sequence data", BMC GENOMICS, BIOMED CENTRAL LTD, LONDON, UK, vol. 12, no. 1, 12 April 2011 (2011-04-12) , page 184, XP021097730, ISSN: 1471-2164, DOI: 10.1186/1471-2164-12-184
- DEREK Y CHIANG ET AL: "High-resolution mapping of copy-number alterations with massively parallel sequencing", NATURE METHODS, vol. 6, no. 1, 30 November 2008 (2008-11-30), pages 99-103, XP055065796, ISSN: 1548-7091, DOI: 10.1038/nmeth.1276
- S. YOON ET AL: "Sensitive and accurate detection of copy number variants using read depth of coverage", GENOME RESEARCH, vol. 19, no. 9, 5 August 2009 (2009-08-05) , pages 1586-1592, XP055167321, ISSN: 1088-9051, DOI: 10.1101/gr.092981.109
- T. CHU ET AL: "Statistical model for whole genome sequencing and its application to minimally invasive diagnosis of fetal genetic disease", BIOINFORMATICS, vol. 25, no. 10, 23 March 2009 (2009-03-23), pages 1244-1250, XP055018601, ISSN: 1367-4803, DOI: 10.1093/bioinformatics/btp156
- CHUNLEI ZHANG ET AL: "A Single Cell Level Based Method for Copy Number Variation Analysis by Low Coverage Massively Parallel Sequencing", PLOS ONE, vol. 8, no. 1, 23 January 2013 (2013-01-23), page e54236, XP55136784, ISSN: 1932-6203, DOI: 10.1371/journal.pone.0054236